Tutorial

The Human Disease Ontology (DO) website navigation





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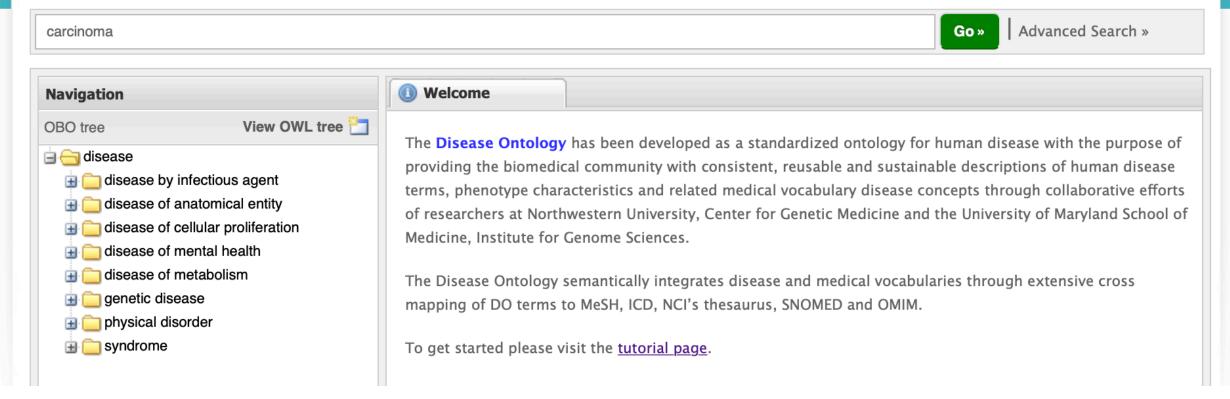
Curation

Downloads

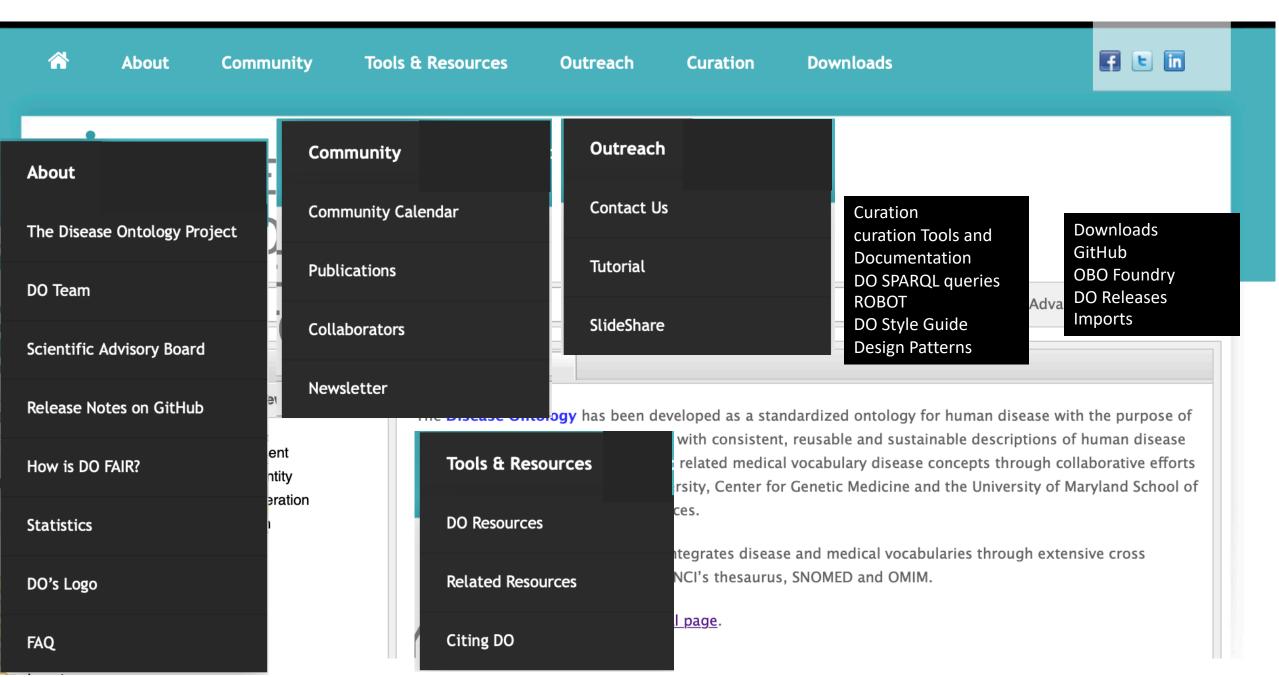




Query disease terms in DO's OBO (default view) or OWL trees. Explore related diseases (e.g. shared anatomy, cell of origin, mode of inheritance, sequence structural variant type, symptom).



Explore the Human Disease Ontology Project Information & Resources



Basic search: text based search of disease

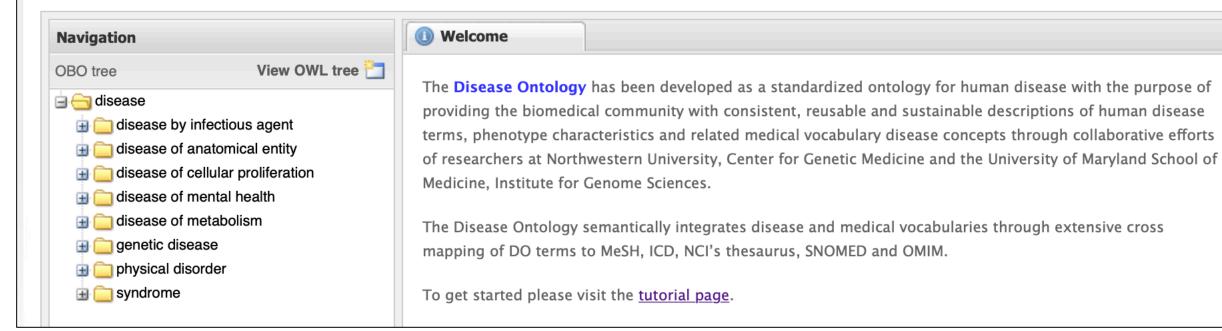
terms



carcinoma

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Advanced Search »

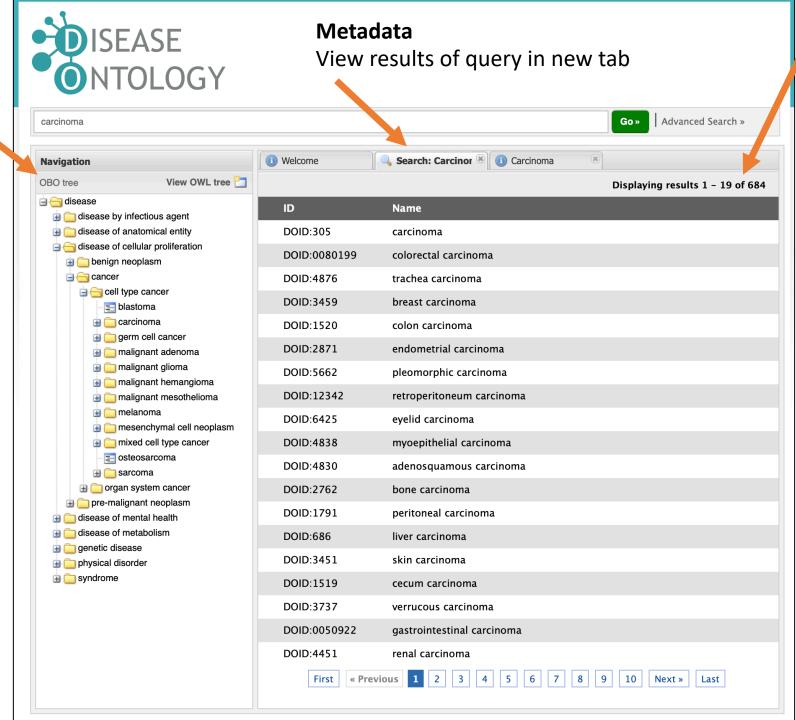


OBO Tree

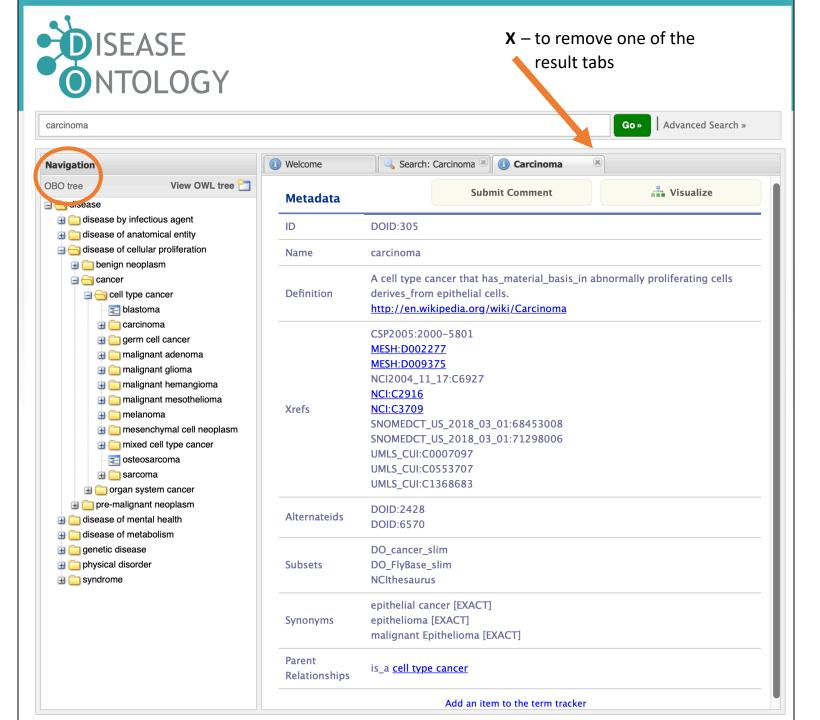
Search doid.obo file

To View Disease MetadataSelect a disease term
or

Term in the OBO Tree



Number of disease terms in query result set



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

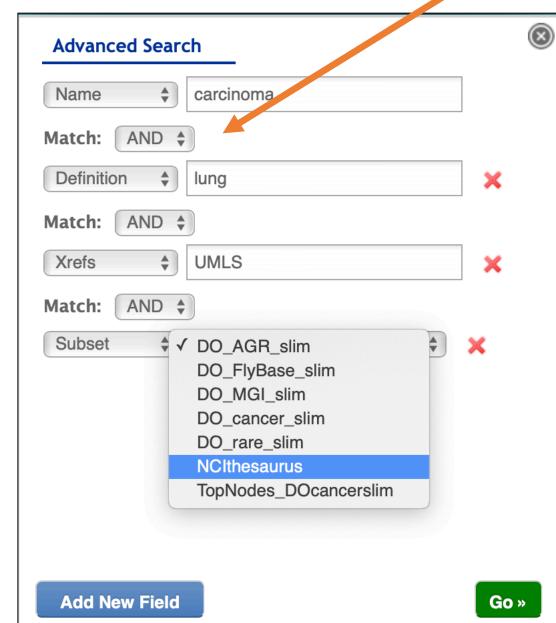
Match: enables Boolean AND, OR or NOT matching across

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.





Search specific metadata fields:

✓ AND OR

NOT

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

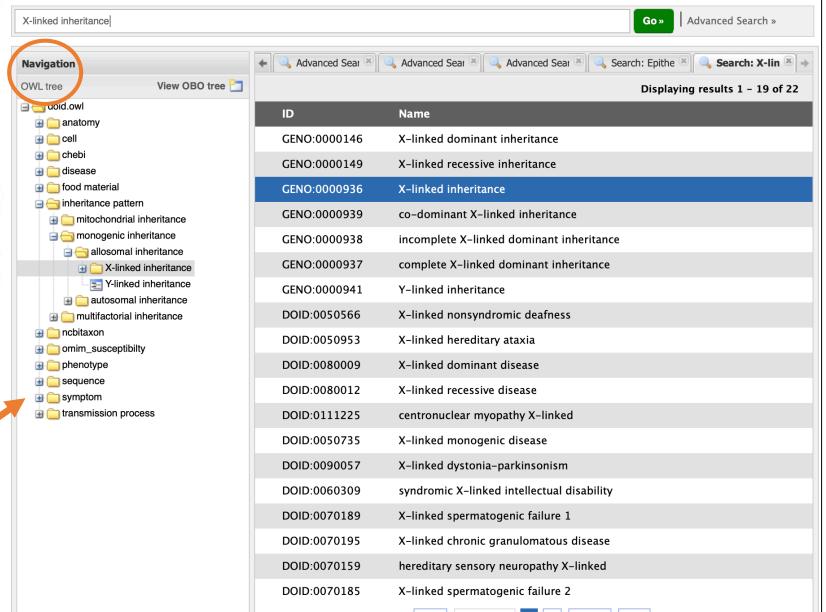
Or

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





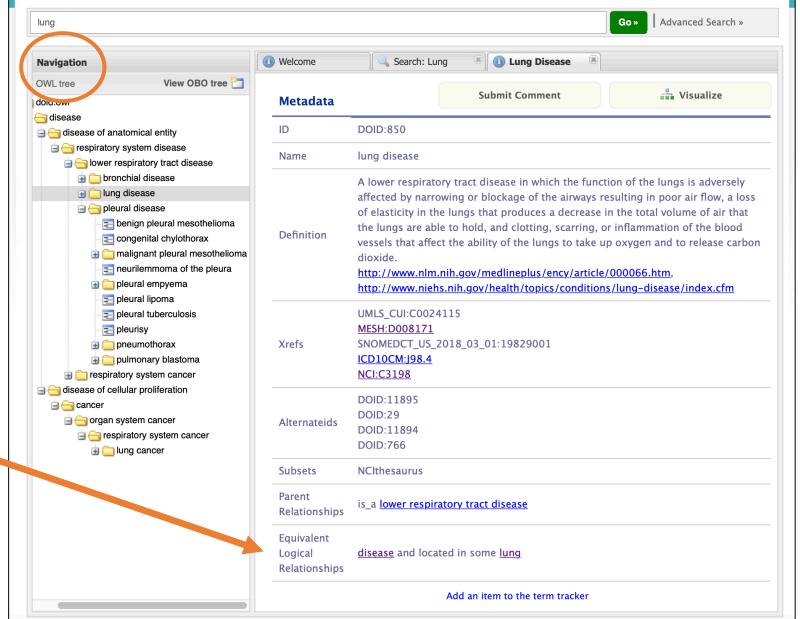
DO OWL Tree

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





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



