MonDO search help

Allowed vocabularies

MonDO (Monarch Disease Ontology) merges several different vocabularies into a single ontology structure, including some non-human disease ontologies. The ClinGen interfaces therefore allow a subset of the merged vocabularies from MonDO to be used in curation. These include:

Vocabulary	Example ID (View in OLS)
Orphanet	Orphanet:93545
Disease Ontology	DOID:9409
NCIt	NCIT:C4089
OMIM	OMIM:100800

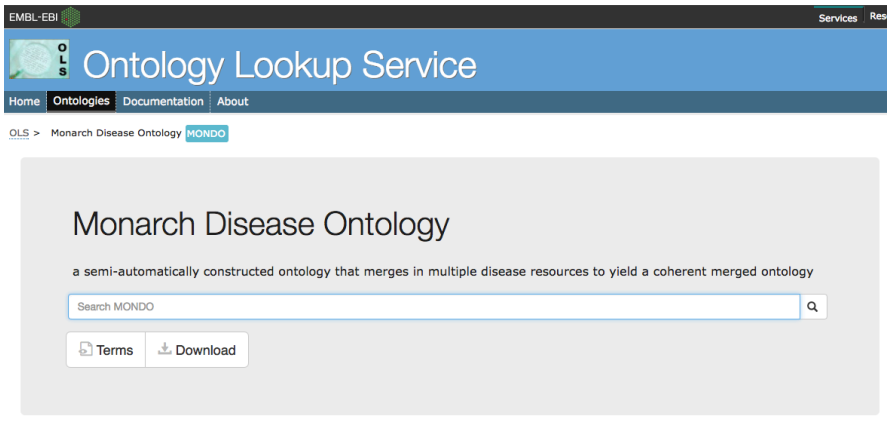
If you click on any of the above examples, you will see them as they are displayed in the Ontology Lookup Service (OLS), which is the resource the interfaces use to return MonDO disease terms via API once you have entered and ID.

Note that all of these have the MonDO label associated with them on the OLS results page– e.g.:

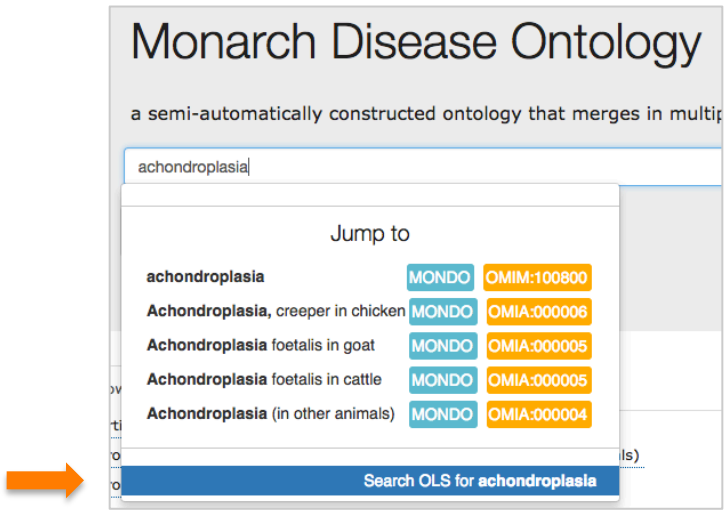
DOID:9409

Finding a MonDO term

1. Use the MonDO OLS Search to find the correct disease term (this search is linked from the disease modal in the ClinGen interfaces).

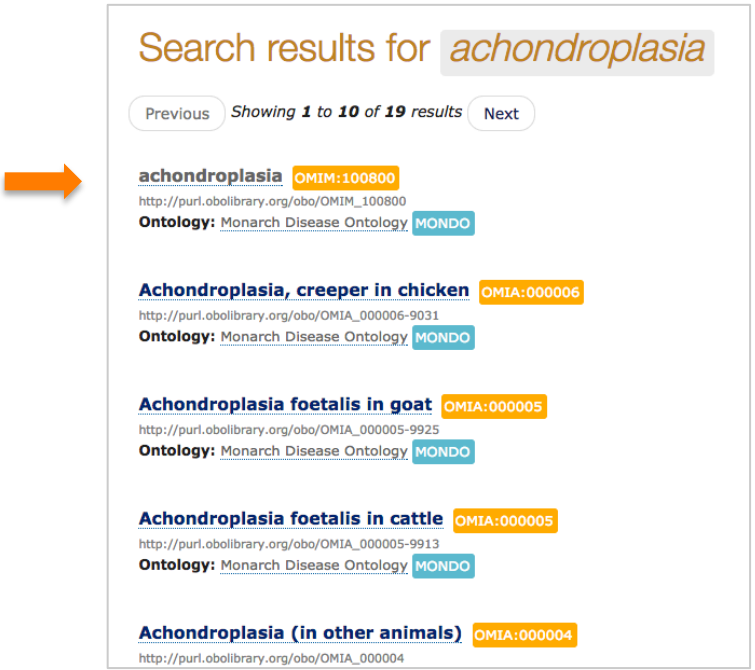


2. The MonDO OLS has a type-ahead function – below is an example of what is found after typing “achondroplasia”:

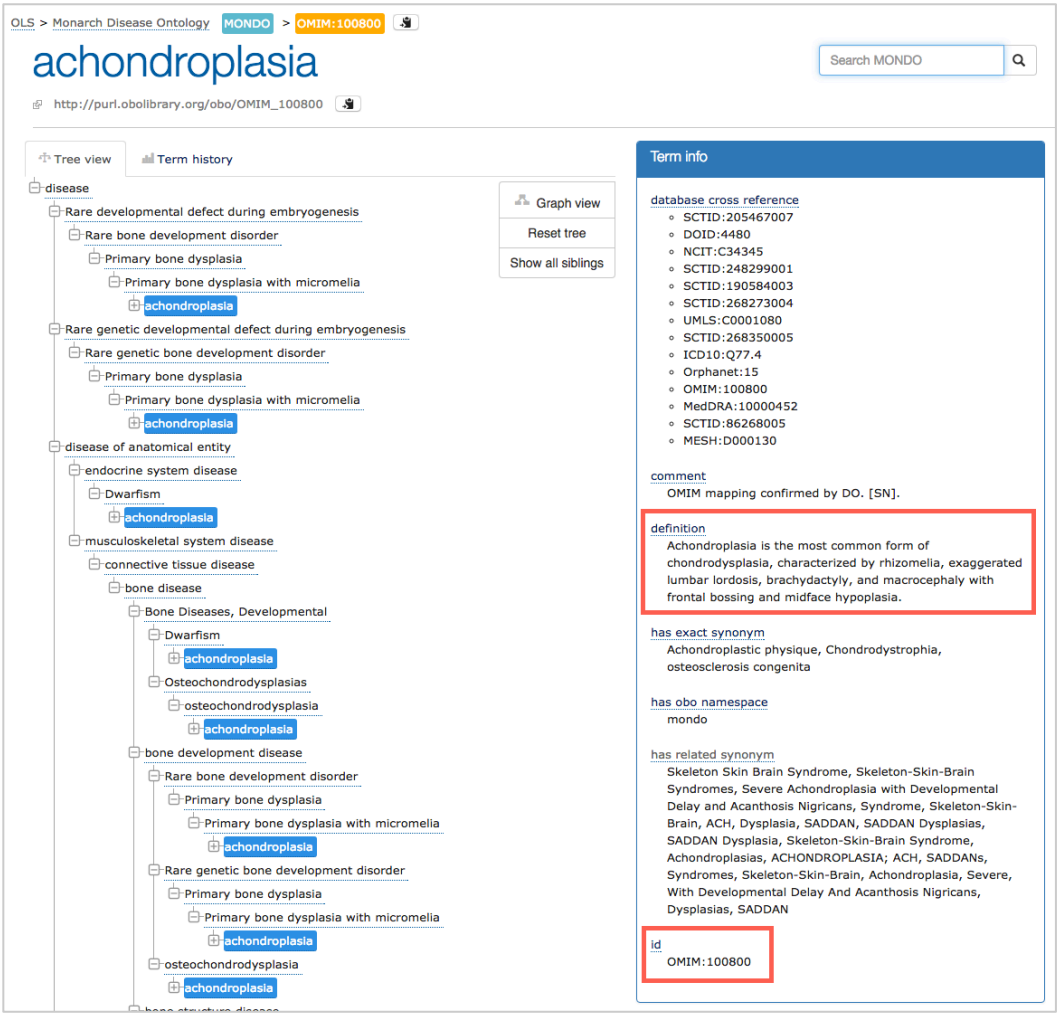


Note that only some of the terms are shown and they may be from vocabularies that are not allowed in the interfaces (e.g. OMIA) – to view all possible terms, click on the dark blue bar (e.g. “Search OLS for achondroplasia”).

After clicking on the blue bar, you should see all MonDO terms associated with your search (these may span several pages in which case you may want to refine your search whenever possible):



3. Click on a term that is one of the 4 MonDO terms allowed (Orphanet, Disease Ontology (DO), NCIt, or OMIM) to check its definition and make certain it is the correct term:



If provided, read the “definition” to make certain it matches the disease to which you are curating.

If the term is correct, copy the “id” in the Term info window (“OMIM:100800” in this example) and paste it into the interface.

Note: the entered ID must begin with “Orphanet:”, “DOID:”, “NCIT:”, or OMIM:”. It must also be a MonDO term, as indicated at the top of the term page:

If you have any questions about searching and identifying MonDO terms using the OLS, please reach out to us at clingen-helpdesk@lists.stanford.edu.