

DO Style Guide The DO Style Guide provides documentation on defining new terms, cleaning up term format, creating definitions, dbxrefs (IDs as references to external resources) and provenance of term definition sources as URLs.

1. Basic guidance

Disease terms should contain: no plurals, lowercase, except for upper cased proper nouns.

New disease terms: a textual definition should be added for each new disease term in the DO. Each textual definition should be provenanced by one or more references (e.g. publications - with their PubMed ID, URL links to authoritative resources, e.g. Genetics Home Reference, NCI thesaurus, Online Mendelian Inheritance in Man (OMIM).

2. Definition Guidelines

The formal definition is stated within the first sentence.

The first sentence always begins with the parent term.

The first sentence should describe the etiology (cause) of the disease.

The second second sentence: additional symptoms, or inheritance information, if needed.

The following sentences may include additional descriptive text.

Example Definitions

A disease that is characterized by X and that involves [Symptoms, Mutations, Transmission]

cardiomyopathy [DOID:0050700]

A heart disease and a myopathy that is characterized by deterioration of the function of the heart muscle.

monogenic disease [DOID:0050177]

A genetic disease that is the result of one or more abnormal alleles and may be dominant (a single copy of the abnormal allele is sufficient to give rise to the disease), semi-dominant, or recessive (requiring both copies of the gene to have an abnormal allele).

Norrie disease [DOID:0060844]

A X-linked recessive disease characterized by degenerative and proliferative changes of the neuroretina resulting in congenital blindness along with progressive mental disorders in about 50% of patients and sensorineural deafness in about 33% of patients, and that has material_basis_in mutation in the NDP gene on chromosome Xp11.

dengue shock syndrome [DOID:0050125]

A dengue disease that involves the most severe form of dengue fever, has_material_basis_in Dengue virus [NCBITaxon:12637] with four serotypes (Dengue virus 1, 2, 3 and 4), which are transmitted_by Aedes mosquito bite. The infection has_symptom easy bruising, has_symptom blood spots, has_symptom bleeding gums, and has_symptom nosebleeds. It is accompanied by circulatory collapse, involves hypotension, narrow pulse pressure (less than or equal to 20mm Hg), or frank shock. The shock occurs after two to six days of symptoms, followed by collapse, weak pulse, and blueness around the mouth.

dementia [DOID:1307]

A cognitive disorder resulting from a loss of brain function affecting memory, thinking, language, judgement and behavior.

striated muscle rhabdoid tumor [DOID:0050924]

A muscle cancer that is located_in striated muscle and has_material_basis_in rhabdoid cells which are large cells with eccentrically located nuclei and abundant, eosinophilic cytoplasm.

3. Obsolete Terms

Some terms in the DO are ‘obsoleted’.

This occurs when:

- Review of terms has concluded that the term does not represent a disease, these may include injuries, symptoms, phenotypes or terms that are no longer considered valid by the research community; OR
- When two DO terms are merged.

When two disease terms are merged, the DOID for the term being merged is added as an alternate id in the second DOID record. The disease name for obsoleted term is prepended with ‘obsolete’.

Example of an obsolete term due to not being a disease

[Term] id: DOID:1000 name: Uterine Rupture synonym: "Rupture of uterus (disorder)" EXACT
[SNOMEDCT_2005_07_31:34430009] synonym: "Ruptured uterus" EXACT
[SNOMEDCT_2005_07_31:156231001] xref: MSH2005_2005_01_17:M0022435 xref:
MSH2006_2005_11_15:D014597

xref: UMLS_CUI:C0042143 xref: UMLS_MSH2005_2005_01_17_AUI:A0113357 xref:
UMLS_MSH2005_2005_01_17_AUI:A0113369 xref:
UMLS_MSH2005_2005_01_17_AUI:A0130889 xref:
UMLS_MSH2005_2005_01_17_AUI:A0130890 xref: UMLS_ST:T047 is_obsolete: true

4. The DO disease names do not include the following phrases utilized in clinical vocabularies:
terms including: with or without; non-specific words: such as may, sometimes, most or typical;
compound terms; other, unspecified, NOS; of other parts; due to other; (disease qualifiers) with
other specified complications;