



The Disease Ontology can be viewed from:

Human Disease Ontology

www.disease-ontology.org

EBI Ontology Lookup Service:

<https://www.ebi.ac.uk/ols/ontologies/doid>

Authoritative Sources for Definitions:

For each definition, include at least once source for the definition.

The sources are added as annotations to the Definition Property, as

'database_cross_reference'

MedlinePlus: <https://medlineplus.gov/braindiseases.html>

Wikipedia: https://en.wikipedia.org/wiki/Central_nervous_system_disease

GeneReviews: <https://www.ncbi.nlm.nih.gov/books/NBK1116/>

Genetics Home Reference: <https://ghr.nlm.nih.gov/condition>

NCI dictionary of cancer terms [\[link\]](#)

Mayo Clinic: <https://www.mayoclinic.org/diseases-conditions/breast-cancer/symptoms-causes/syc-20352470>

PubMed ID <https://www.ncbi.nlm.nih.gov/pubmed/>

Format examples:

https://en.wikipedia.org/wiki/NUT_midline_carcinoma

<https://omim.org/entry/608749>

<https://www.ncbi.nlm.nih.gov/pubmed/21221870>

Definitions identify the parent term and specify what is distinct about each child term. The source(s) of knowledge/information for each definition is records as an 'xref', by the URL of the source Information.

- (1) Anatomical location (Tissue of Origin):
 - defined by ontology relation: `located_in` [anatomy]
 - linked to anatomy ontology ([UBERON](#))

- (2) Cell of Origin
 - defined by ontology relation: `has_material_basis_in` [tissue of origin]
 - defined by ontology relation: `derives_from` [cell of origin]
 - linked to the [Cell ontology](#)

cell type cancer:

Example: **blastoma**: A cell type cancer that `has_material_basis_in` abnormally proliferating cells `derives_from` precursor cells called blast cells.

organ system cancer:

bladder carcinoma

A urinary bladder cancer that `has_material_basis_in` abnormally proliferating cells `derives_from` epithelial cells.

NCI dictionary of cancer terms [\[link\]](#)

<u>cell type cancer</u>	<u>definition text</u>
adenocarcinoma	<code>has_material_basis_in</code> abnormally proliferating cells, <code>derives_from</code> epithelial cells, which originate in glandular tissue.
blastoma	<code>has_material_basis_in</code> abnormally proliferating cells <code>derives_from</code> precursor cells called blast cells.
carcinoma	<code>has_material_basis_in</code> abnormally proliferating cells <code>derives_from</code> epithelial cells.
germ cell cancer	<code>has_material_basis_in</code> abnormally proliferating cells <code>derives_from</code> germ cells.
glioma	<code>has_material_basis_in</code> glial cells and is <code>located_in</code> brain or <code>located_in</code> spine.
hemangioma	A cell type cancer of vascular origin, characterized by proliferation of endothelial cells in and about the vascular lumen.
mesothelioma	<code>has_material_basis_in</code> mesothelial tissue.
melanoma	<code>has_material_basis_in</code> abnormally proliferating cells <code>derives_from</code> melanocytes which are found in skin, the bowel and the eye.
mesenchymal cell neoplasm	<code>has_material_basis_in</code> abnormally proliferating cells <code>derives_from</code> embryonic connective tissue
mixed cell type cancer	<code>has_material_basis_in</code> abnormally proliferating cells <code>derives_from</code> two germinal layers of tissue
sarcoma	<code>has_material_basis_in</code> abnormally proliferating cells <code>derives_from</code> embryonic mesoderm