Supplemental Document #3

Comorbidity Diseases Information in Disease Ontology

In this supplemental document, the comorbidity diseases information in the disease ontology are described.

No.	Disease	Information in DO
1.	Nervous System Disease	**ID** DOID:863 **Definition** A disease of anatomical entity that is located_in the central nervous system or located_in the peripheral nervous system. http://en.wikipedia.org/wiki/Nervous_system. **Xrefs** ICD10CM:G98 ICD9CM:349.9 MESH:D009422 NCI:C26835 SNOMEDCT_US_2023_03_01:155262005 UMLS_CUI:C0027765 **Subsets** DO_CFDE_slim DO_GXD_slim DO_MGI_slim NCIthesaurus **Parent Relationships** is a disease of anatomical entity
2.	Cardiovascular System Disease	**ID** DOID:1287
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2.	Cardiovascular System Disease	**Definition** A disease of anatomical entity which occurs in the blood, heart, blood vessels or the lymphatic system that passes nutrients (such as amino acids and electrolytes), gases, hormones, blood cells or lymph to and from cells in the body to help fight diseases and help stabilize body temperature and pH to maintain homeostasis. http://en.wikipedia.org/wiki/Circulatory_system. **Xrefs** ICD9CM:429.2 MESH:D002318 NCI:C2931 SNOMEDCT_US_2023_03_01:266275004 UMLS_CUI:C0007222 **Alternate IDs** DOID:73 **Subsets** DO_AGR_slim DO_CFDE_slim DO_GXD_slim DO_MGI_slim DO_RAD_slim NCIthesaurus **Synonyms** disease of subdivision of hemolymphoid system [EXACT] **Parent Relationships** is a disease of anatomical entity
3.	Gastrointestinal System Disease	**ID** DOID:77
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3.	Gastrointestinal System Disease	**Definition** A disease of anatomical entity that is located_in the gastrointestinal tract. http://en.wikipedia.org/wiki/Human_gastrointestinal_tract. **Xrefs** ICD10CM:K92.9 ICD9CM:520-579.99 MESH:D004066 SNOMEDCT_US_2023_03_01:53619000 UMLS_CUI:C0012242 **Alternate IDs** DOID:27 DOID:944 **Subsets** DO_AGR_slim DO_CFDE_slim DO_GXD_slim DO_MGI_slim **Synonyms** alimentary system disease [EXACT] digestive system disorder [EXACT] Gastroenteropathy [EXACT] gastrointestinal disease [EXACT] gastrointestinal disorder [EXACT] GIT disease [EXACT] **Parent Relationships** is a disease of anatomical entity

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4.	Disease of Mental Health	**ID** DOID:150 **Definition** A disease that involves a psychological or behavioral pattern generally associated with subjective distress or disability that occurs in an individual, and which are not a part of normal development or culture. http://en.wikipedia.org/wiki/Mental_disorder. **Xrefs** ICD10CM:F99 MESH:D001523 NCI:C2893 SNOMEDCT_US_2023_03_01:74732009 UMLS_CUI:C0004936 **Subsets** DO_AGR_slim DO_CFDE_slim DO_GXD_slim DO_MGI_slim DO_RAD_slim NCIthesaurus **Parent Relationships** is a disease
5.	Retinal Degeneration	**ID** DOID:8466 **Definition** A retinal disease that is characterized by deterioration of the retina caused by the progressive and eventual death of the cells of the retina. https://en.wikipedia.org/wiki/Retinal_degeneration_(rhodopsin_mutation).
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5.	Retinal Degeneration	**Xrefs** MESH:D012162 NCI:C34979 SNOMEDCT_US_2023_03_01:95695004 UMLS_CUI:C0035304 **Subsets** DO_FlyBase_slim NCIthesaurus **Synonyms** degeneration of retina [EXACT] **Parent Relationships** is a eye degenerative disease is a retinal disease
6.	Lung Disease	**ID** DOID:850 **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. http://www.niehs.nih.gov/health/topics/conditions/lung-disease/index.cfm, http://www.nlm.nih.gov/medlineplus/ency/article/000066.htm.

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6.	Lung Disease	**Xrefs** ICD10CM:J98.4 MESH:D008171 NCI:C3198 SNOMEDCT_US_2023_03_01:266374002 UMLS_CUI:C0024115 **Alternate IDs** DOID:11894 DOID:11895 DOID:29 DOID:766 **Subsets** DO_RAD_slim NCIthesaurus **Parent Relationships** is a lower respiratory tract disease
7.	Lower Respiratory Tract Disease	**ID** DOID:0050161 **Definition** A respiratory system disease which involves the lower respiratory tract. http://en.wikipedia.org/wiki/lower_respiratory_tract. **Xrefs** ICD9CM:478.19 SNOMEDCT_US_2023_03_01:195823002 UMLS_CUI:C0029581

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7.	Lower Respiratory Tract Disease	**Subsets** DO_RAD_slim **Parent Relationships** is a respiratory system disease
8.	Autoimmune Disease	**ID** DOID:417 **Definition** An immune system disease that is an overactive immune response of the body against substances and tissues normally present in the body resulting from an abnormal functioning of the immune system that results in the production of antibodies or T cell directed against the host tissues. http://en.wikipedia.org/wiki/Autoimmune_disease. **Xrefs** ICD9CM:720 OMIM:109100 UMLS_CUI:C0003089 **Synonyms** autoimmune hypersensitivity disease [EXACT] hypersensitivity reaction type II disease [EXACT] **Parent Relationships** is a primary immunodeficiency disease
9.	Dermatitis	**ID** DOID:2723 **Definition** A skin disease characterized by itchy, erythematous, vesicular, weeping and crusting patches of skin. http://en.wikipedia.org/wiki/Dermatitis, http://www.nlm.nih.gov/medlineplus/eczema.html.

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9.	Dermatitis	**Xrefs** ICD10CM:L30.9 MESH:D003872 NCI:C2983 SNOMEDCT_US_2023_03_01:238538009 UMLS_CUI:C0011603 **Alternate IDs** DOID:8614 DOID:8917 **Subsets** NCIthesaurus **Synonyms** eczema [EXACT] skin inflammation [EXACT] **Parent Relationships** is a skin disease
10.	Bone Disease	**ID** DOID:0080001 **Definition** A connective tissue disease that affects the structure or development of bone or causes an impairment of normal bone function. http://en.wikipedia.org/wiki/Bone_disease. **Xrefs** ICD10CM:M89.9 MESH:D001847 SNOMEDCT_US_2023_03_01:76069003 UMLS_CUI:C0005940

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10.	Bone Disease	**Alternate IDs** DOID:1290 **Synonyms** skeletal disease [RELATED] **Parent Relationships** is a connective tissue disease
11.	Kidney Disease	**ID** DOID:557 **Definition** A urinary system disease that is located_in the kidney. http://www.nlm.nih.gov/medlineplus/kidneydiseases.html. **Xrefs** EFO:0003086 ICD10CM:N08 MESH:D007674 NCI:C3149 SNOMEDCT_US_2023_03_01:266612003 UMLS_CUI:C0022658 **Alternate IDs** DOID:11705 **Subsets** DO_FlyBase_slim DO_RAD_slim NCIthesaurus

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11.	Kidney Disease	**Synonyms** impaired renal function disease [EXACT] nephropathy [EXACT] **Parent Relationships** is a urinary system disease
12.	Autosomal Dominant Disease	**ID** DOID:0050736 **Definition** An autosomal genetic disease that is characterized by the presence of one disease-associated mutation of a gene which is sufficient to cause the disease. http://ghr.nlm.nih.gov/glossary=autosomaldominant, http://ghr.nlm.nih.gov/handbook/inheritance/inheritancepatterns, http://www.nlm.nih.gov/medlineplus/ency/article/002049.htm. **Parent Relationships** is a autosomal genetic disease
13.	Thyroid Gland Disease	**ID** DOID:50 **Definition** An endocrine system disease that is located_in the thyroid. http://en.wikipedia.org/wiki/Thyroid. **Xrefs** ICD10CM:E07.9 ICD9CM:246.9 MESH:D013959 NCI:C26893 SNOMEDCT_US_2023_03_01:14304000 UMLS_CUI:C0040128

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13.	Thyroid Gland Disease	**Subsets** NCIthesaurus **Parent Relationships** is a endocrine system disease
14.	Genetic Disease	**ID** DOID:630 **Definition** A disease that has_material_basis_in genetic variations in the human genome. http://ghr.nlm.nih.gov/. **Xrefs** MESH:D030342 NCI:C3101 SNOMEDCT_US_2023_03_01:32895009 UMLS_CUI:C0019247 **Subsets** DO_CFDE_slim DO_RAD_slim NCIthesaurus **Parent Relationships** is a disease
15.	Myositis	**ID** DOID:633 **Definition** A myopathy characterized by muscle inflammation. http://www.nlm.nih.gov/medlineplus/myositis.html, https://en.wikipedia.org/wiki/Myositis.

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15.	Myositis	**Xrefs** ICD10CM:M60 MESH:D009220 NCI:C27578 OMIM:160750 SNOMEDCT_US_2023_03_01:26889001 UMLS_CUI:C0027121 **Subsets** NCIthesaurus **Synonyms** Inflammatory disorder of muscle [EXACT] **Parent Relationships** is a myopathy
16.	Branch Retinal Artery Occlusion	**ID** DOID:13094 **Xrefs** ICD10CM:H34.23 ICD9CM:362.32 MESH:D015356 NCI:C34436 SNOMEDCT_US_2023_03_01:50821009 UMLS_CUI:C0006123 **Subsets** NCIthesaurus **Synonyms** Arterial retinal branch occlusion [EXACT] retinal arterial branch occlusion [EXACT]

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16.	Branch Retinal Artery Occlusion	**Parent Relationships** is a retinal artery occlusion
17.	Retinal Vein Occlusion	**ID** DOID:1727 **Xrefs** MESH:D012170 NCI:C34981 SNOMEDCT_US_2023_03_01:46085004 UMLS_CUI:C0035328 **Subsets** NCIthesaurus **Synonyms** Occlusion, of retinal vein [EXACT] **Parent Relationships** is a vein disease is a retinal vascular occlusion
18.	Central Retinal Artery Occlusion	**ID** DOID:13098 **Definition** A retinal artery occlusion characterized by blockage of blood flow through the central retinal artery. http://en.wikipedia.org/wiki/Central_retinal_artery_occlusion.
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18.	Central Retinal Artery Occlusion	**Xrefs** ICD10CM:H34.1 ICD9CM:362.31 MESH:D015356 NCI:C34456 SNOMEDCT_US_2023_03_01:38742007 UMLS_CUI:C0007688 **Subsets** NCIthesaurus **Parent Relationships** is a retinal artery occlusion
19.	Retinal Artery Occlusion	**ID** DOID:8483 **Xrefs** MESH:D015356 NCI:C34978 SNOMEDCT_US_2023_03_01:232035005 UMLS_CUI:C0035302 **Subsets** NCIthesaurus **Parent Relationships** is a retinal vascular occlusion is a artery disease
20.	Developmental Disorder of Mental Health	**ID** DOID:0060037

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20.	Developmental Disorder of Mental Health	**Definition** A disease of mental health that occur during a child's developmental period between birth and age 18 resulting in retarding of the child's psychological or physical development. http://en.wikipedia.org/wiki/Developmental_disorders. **Subsets** DO_RAD_slim **Parent Relationships** is a disease of mental health
21.	Cognitive Disorder	**ID** DOID:1561 **Definition** A disease of mental health that affects cognitive functions including memory processing, perception and problem solving. http://en.wikipedia.org/wiki/Cognitive_disorder. **Xrefs** ICD10CM:F09 MESH:D019965 NCI:C34870 SNOMEDCT_US_2023_03_01:111479008 UMLS_CUI:C0029227 **Subsets** DO_RAD_slim NCIthesaurus **Synonyms** cognitive disease [EXACT] Organic Mental disorder [RELATED]

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21.	Cognitive Disorder	**Parent Relationships** is a disease of mental health
22.	Primary Immunodeficiency Disease	**ID** DOID:612 **Definition** An immune system disease that results when one or more essential parts of the immune system is missing or not working properly at birth due to a genetic mutation. http://www.nichd.nih.gov/publications/pubs/primary_immuno.cfm#PrimaryImmunoDiseases. **Xrefs** ICD10CM:D84.9 ICD9CM:279.3 KEGG:05340 MESH:D007153 NCI:C39725 OMIM:242850 OMIM:PS300755 SNOMEDCT_US_2023_03_01:191005003 UMLS_CUI:C0021051 **Subsets** DO_FlyBase_slim NCIthesaurus **Synonyms** hypoimmunity [EXACT] immune deficiency disorder [EXACT] immunodeficiency syndrome [EXACT] **Parent Relationships** is a immune system disease

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23.	Hypersensitivity Reaction Disease	**ID** DOID:0060056 **Definition** An immune system disease that has_material_basis_in abnormal immune responses. http://en.wikipedia.org/wiki/Hypersensitivity, http://www.ncbi.nlm.nih.gov/books/NBK27136/. **Parent Relationships** is a immune system disease
24.	Bile Duct Disease	**ID** DOID:4138 **Definition** A biliary tract disease located_in one or more bile ducts. https://medlineplus.gov/bileductdiseases.html, https://www.health.harvard.edu/a_to_z/bile-duct-diseases-a-to-z, https://www.nyp.org/cadc/liver-diseases-and-transplantation/bile-duct-disorders. **Xrefs** MESH:D001649 NCI:C96716 SNOMEDCT_US_2023_03_01:118926004 UMLS_CUI:C0005395 **Subsets** NCIthesaurus **Synonyms** bile duct disorder [EXACT] disorder of bile duct [EXACT]

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24.	Bile Duct Disease	**Parent Relationships** is a biliary tract disease
25.	Polyneuropathy	**ID** DOID:1389 **Definition** A peripheral system disease that is characterized by damage affecting peripheral nerves (peripheral neuropathy) in roughly the same areas on both sides of the body, featuring weakness, numbness, pins-and-needles, and burning pain. https://en.wikipedia.org/wiki/Polyneuropathy, https://www.virginiamason.org/polyneuropathy. **Xrefs** ICD10CM:A69.22 MESH:D011115 NCI:C26951 SNOMEDCT_US_2023_03_01:193166009 UMLS_CUI:C0152025 **Subsets** DO_FlyBase_slim NCIthesaurus **Parent Relationships** is a peripheral nervous system disease
26.	Cauda Equina Syndrome	**ID** DOID:11577 **Definition** A peripheral nervous system disease that involves an acute loss of function of the lumbar plexus, neurologic elements (nerve roots) of the spinal canal below the termination (conus) of the spinal cord. http://en.wikipedia.org/wiki/Cauda_equina_syndrome.

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26.	Cauda Equina Syndrome	**Xrefs** GARD:10987 ICD10CM:G83.4 ICD9CM:344.6 MESH:D000077684 NCI:C35436 SNOMEDCT_US_2023_03_01:89356000 UMLS_CUI:C0392548 **Subsets** DO_rare_slim NCIthesaurus **Parent Relationships** is a peripheral nervous system disease is a syndrome
27.	Adrenoleukodystrophy	**ID** DOID:10588 **Definition** A leukodystrophy that disrupts the breakdown of very-long-chain fatty acids resulting in progressive brain damage, failure of the adrenal glands and eventually death. http://en.wikipedia.org/wiki/Adrenoleukodystrophy, https://ghr.nlm.nih.gov/condition/x-linked-adrenoleukodystrophy. **Xrefs** ICD10CM:E71.52 MESH:D000326 NCI:C61252 OMIM:300100 SNOMEDCT_US_2023_03_01:65389002 UMLS_CUI:C0162309

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27.	Adrenoleukodystrophy	**Alternate IDs** DOID: 13451 **Subsets** NCIthesaurus **Synonyms** ALD [EXACT] Bronze Schilder disease [EXACT] diffuse sclerosis [EXACT] Encephalitis periaxialis concentrica [EXACT] Encephalitis periaxialis, Schilder's [EXACT] Siemerling-Creutzfeldt Disease [EXACT] sudanophilic cerebral sclerosis [EXACT] X-linked adrenoleukodystrophy [EXACT] **Parent Relationships** is a leukodystrophy is a X-linked recessive disease
28.	Hydrocephalus	**ID** DOID:10908 **Definition** A cerebral degeneration characterized by an abnormal accumulation of cerebrospinal fluid in the ventricles of the brain, leading to progressive enlargement of the head. http://en.wikipedia.org/wiki/Hydrocephalus, http://ghr.nlm.nih.gov/glossary=hydrocephalus, http://www.mayoclinic.org/diseases-conditions/hydrocephalus/basics/definition/con-20030706?_ga=1.124310025.2017809229.1415219956.

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28.	Hydrocephalus	**Xrefs** GARD:6682 ICD10CM:G91 MESH:D006849 NCI:C3111 OMIM:123155 OMIM:236600 OMIM:236635 OMIM:307000 OMIM:615219 ORDO:2182 ORDO:2185 SNOMEDCT_US_2023_03_01:267687006 UMLS_CUI:C0020255 **Subsets** DO_rare_slim NCIthesaurus **Synonyms** hydrocephalus, nonsyndromic, autosomal recessive [EXACT] hydrocephalus, X-linked [EXACT] **Parent Relationships** is a cerebral degeneration
29.	Ataxia with Oculomotor Apraxia Type 1	**ID** DOID:0050754

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29.	Ataxia with Oculomotor Apraxia Type 1	**Definition** An autosomal recessive cerebellar ataxia that is characterized by progressive cerebellar ataxia including oculomotor apraxia, severe neuropathy and hypoalbuminemia, has_material_basis_in autosomal recessive inheritance of mutation in the APTX gene. https://rarediseases.info.nih.gov/diseases/9283/ataxia-oculomotor-apraxia-type-1. **Xrefs** GARD:9283 OMIM:208920 **Subsets** DO_rare_slim **Parent Relationships** is a autosomal recessive cerebellar ataxia
30.	Horner's Syndrome	**ID** DOID:11486 **Definition** An autonomic neuropathy that is characterized by the classic triad of unilateral ptosis, unilateral miosis with anisocoria, and ipsilateral facial anhidrosis, resulting from unilateral paralysis of the cervical sympathetics. https://pubmed.ncbi.nlm.nih.gov/14610154/. **Xrefs** GARD:6670 ICD10CM:G90.2 MESH:D006732 NCI:C28155 OMIM:143000 SNOMEDCT_US_2023_03_01:192922002 UMLS_CUI:C0019937

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30.	Horner's Syndrome	**Subsets** DO_rare_slim NCIthesaurus **Synonyms** Bernard Horner syndrome [EXACT] cervical sympathetic paralysis [EXACT] Horner syndrome [EXACT] **Parent Relationships** is a autonomic neuropathy
31.	Polyp of Middle Ear	**ID** DOID:7439 **Xrefs** ICD10CM:H74.4 NCI:C6933 SNOMEDCT_US_2023_03_01:155244001 UMLS_CUI:C0271466 **Subsets** NCIthesaurus **Synonyms** polyp - middle ear [EXACT] polyp of the middle ear [EXACT] **Parent Relationships** is a middle ear disease
32.	Bell's Palsy	**ID** DOID:12506

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32.	Bell’s Palsy	**Definition** A facial paralysis resulting from dysfunction in the cranial nerve VII (facial nerve). http://en.wikipedia.org/wiki/Bell%27s_palsy. **Xrefs** GARD:5906 ICD10CM:G51.0 ICD9CM:351.0 MESH:D020330 SNOMEDCT_US_2023_03_01:193093009 UMLS_CUI:C0376175 **Subsets** DO_rare_slim **Synonyms** Bell palsy [EXACT] Bell's (facial) palsy [EXACT] **Parent Relationships** is a facial paralysis