

2015 Disease Ontology update: DO's expanded curation activities to connect disease- related data

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Biocuration 2015

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ICD - 9



NCIthesaurus
































 DISEASE ONTOLOGY

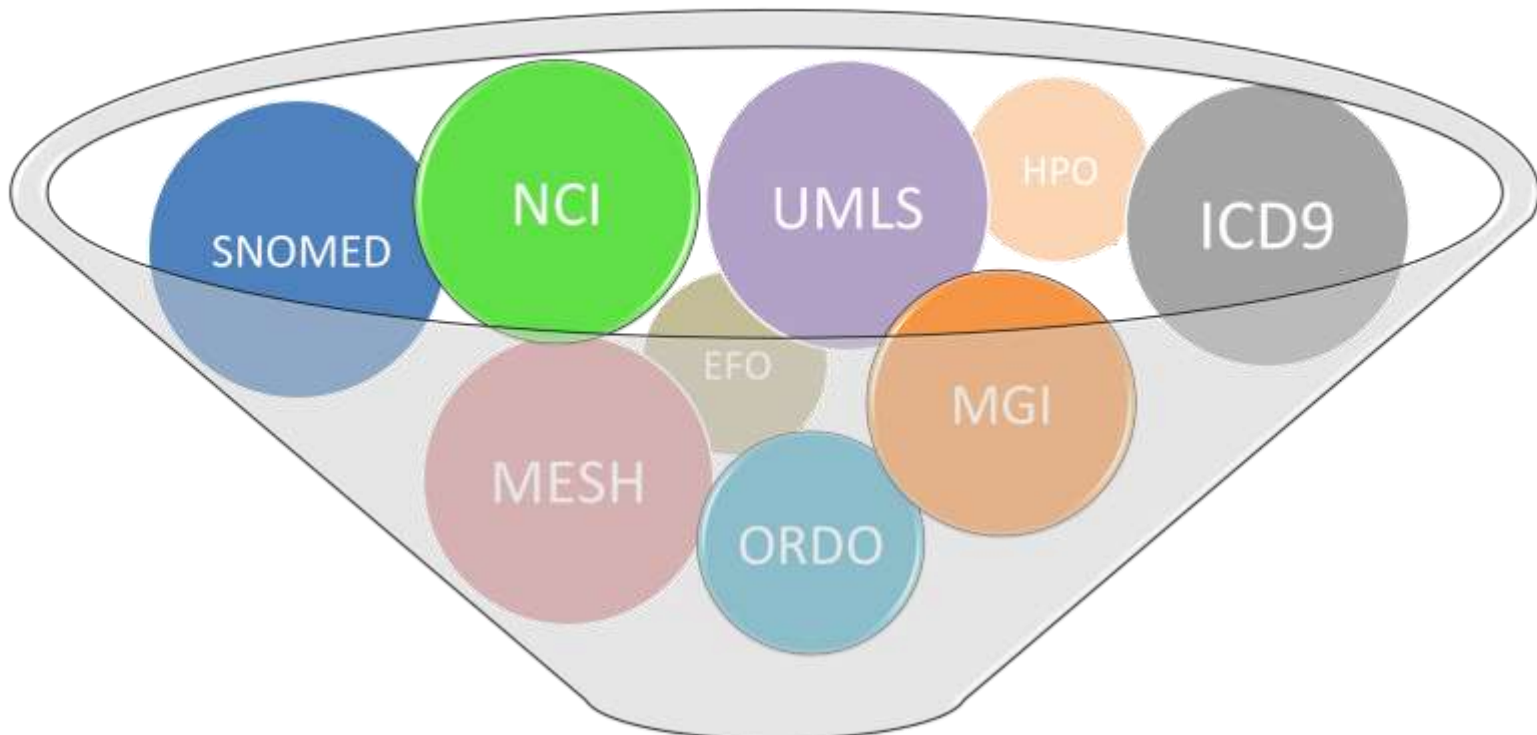
NCBI
NEW MeSH
NLM Controlled Vocabulary

OMIM
Online Mendelian Inheritance in Man
 Johns Hopkins University



- ▶  disease
 - ▶  disease by infectious agent
 - ▶  disease of anatomical entity
 - ▶  disease of cellular proliferation
 - ▶  disease of mental health
 - ▶  disease of metabolism
 - ▶  genetic disease
 - ▶  physical disorder
 - ▶  syndrome

- ▶  disease
 - ▶  disease by infectious agent
 - ▶  bacterial infectious disease
 - ▶  fungal infectious disease
 - ▶  parasitic infectious disease
 - ▶  viral infectious disease
 - ▶  disease of anatomical entity
 - ▶  cardiovascular system disease
 - ▶  endocrine system disease
 - ▶  gastrointestinal system disease
 - ▶  immune system disease
 - ▶  integumentary system disease
 - ▶  musculoskeletal system disease
 - ▶  connective tissue disease
 - ▶  muscular disease
 - ▶  nervous system disease
 - ▶  reproductive system disease
 - ▶  respiratory system disease
 - ▶  thoracic disease
 - ▶  urinary system disease
 - ▶  disease of cellular proliferation
 - ▶  benign neoplasm
 - ▶  cancer
 - ▶  pre-malignant neoplasm
 - ▶  disease of mental health
 - ▶  disease of metabolism
 - ▶  acquired metabolic disease
 - ▶  inherited metabolic disorder
 - ▶  genetic disease
 - ▶  physical disorder
 - ▶  syndrome



DISEASE ONTOLOGY

DO_cancer_slim

Go »

Advanced Search »

Navigation

Open new metadata panel

disease

- ▶ disease by infectious agent
- ▶ disease of anatomical entity
- ▶ disease of cellular proliferation
- ▶ benign neoplasm
- ▶ cancer
 - ▶ cell type cancer
 - ▶ organ system cancer
 - ▶ cardiovascular cancer
 - ▶ endocrine gland cancer
 - ▶ adrenal gland cancer
 - ▶ adrenal carcinoma
 - ▶ adrenal cortex cancer
 - ▶ adrenocortical carcinoma
 - ▶ adrenal gland ganglioneurob
 - ▶ adrenal medulla cancer
 - ▶ adrenal neuroblastoma
 - ▶ pancreatic cancer
 - ▶ parathyroid carcinoma
 - ▶ pineal gland cancer
 - ▶ pituitary cancer
 - ▶ thyroid cancer
 - ▶ gastrointestinal system cancer
 - ▶ immune system cancer
 - ▶ integumentary system cancer

Welcome

Search: DO_cance

Adrenocortical C

Metadata

Visualize

DOID	DOID:3948
Name	adrenocortical carcinoma
Definition	An adrenal cortex cancer that forms in the outer layer of tissue of the adrenal gland and that has_material_basis_in abnormally proliferating cells derives_from epithelial cells. http://www.cancer.gov/cancertopics/types/adrenocortical
Xrefs	MSH:D018268 NCI:C9325 OMIM:202300 SNOMEDCT_2010_1_31:2227007 SNOMEDCT_2010_1_31:255035007 UMLS_CUI:C0206686
Subsets	DO_cancer_slim
Synonyms	Adrenal cortical carcinoma (morphologic abnormality) [EXACT] carcinoma of the Adrenal cortex [EXACT]
Relationships	is_a adrenal cortex cancer

Add an item to the term tracker

	MeSH	OMIM	SNOMED-CT	UMLS direct	UMLS indirect	Total
DO	2 908	1 864	13 276	12 170	8 354	35 895
ORDO	1 705	5 201	1 943	7 997	2 285	16 920
CTD	11 332	3 977	0	0	6 650	21 959
NDF-RT	4 661	0	0	4 699	7 922	12 906
KEGG	1 295	2 690	0	0	4 482	8 467
HPO	0	6 528	0	0	8 000	14 528
NCI	0	0	0	3 910	0	3 910

DO

Terms: 8,946
Synonyms: 16,023
Xrefs: 38,865

**Top Nodes
DO -Cancer Slim**

DO-MGI Slim

Terms: 400
Synonyms: 744
Xrefs: 4619

Terms: 73
Synonyms: 348
Xrefs: 597

Terms: 187
Synonyms: 418
Xrefs: 802

DO-Cancer Slim

Genes identified as potential models

Nhx-5, nlg-1, shn-1
C. elegans neuroligin homolog
WBGene00006412

AUTISM

[autism spectrum disorder]

DOID:12849 autistic disorder
OMIM: 209850



FlyBase

model of autism spectrum disorder

FlyBase allele: Dmel\Nlg2^{KO17},

gene: Neuroligin 2

FBgn0031866

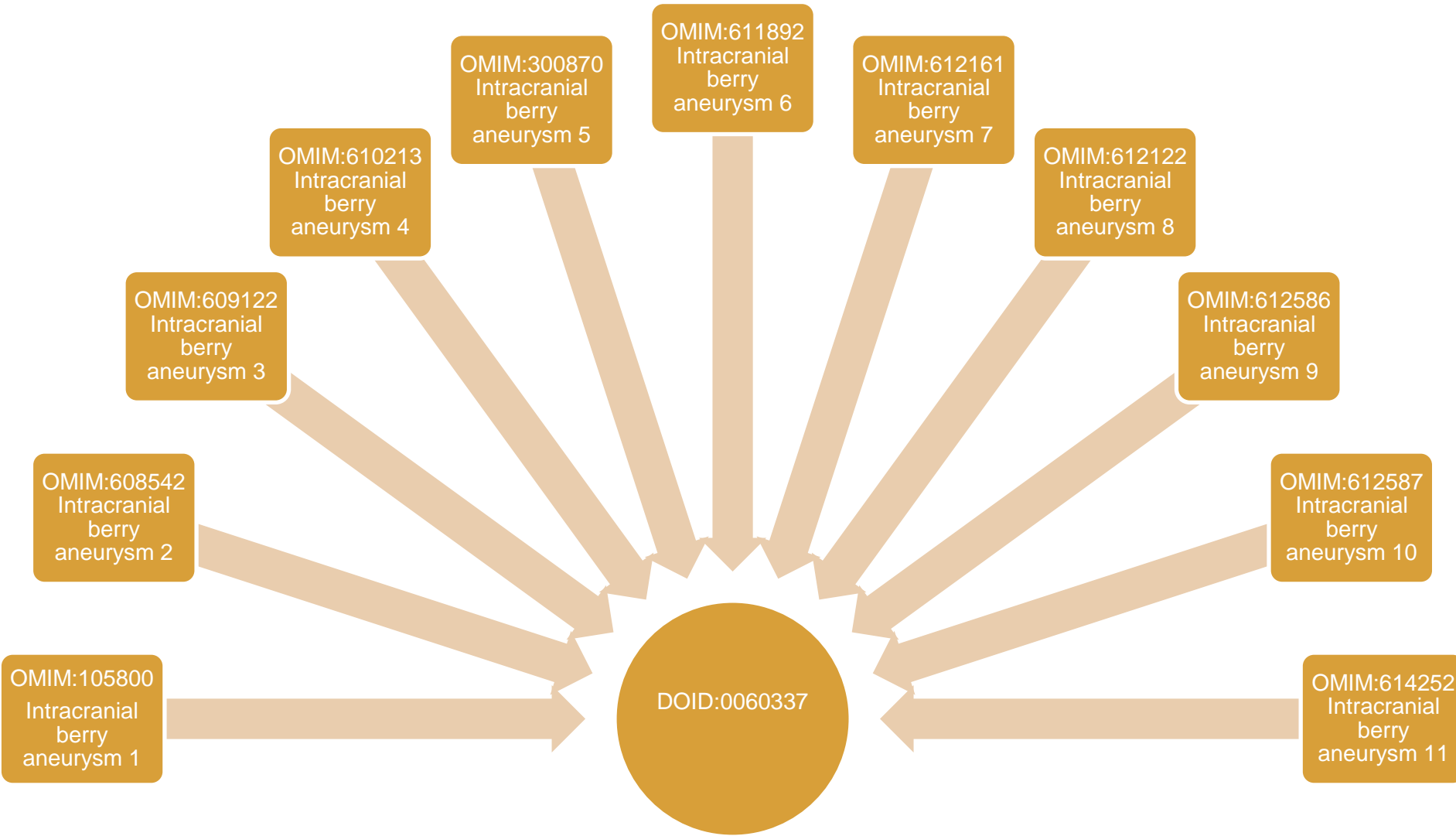


32 mouse models

Nlgn4|Gt(XST093)Byg

Gene: neuroligin 4-like

MGI:3775191



DO Community Projects

DO Disease Mappings

MGI 3434 MGI disease MODs
Reactome 770 + terms mapped
PRO 50 unique MIM – DO terms in PRO

FlyBase (alleles): 2,289 Fbal_ID-DO mappings
annotating disease models

SGD: 1340 diseases

Experimental Factor Ontology (EFO): 137 DO — EFO mappings

Cancer Variant Projects

Clinical Genomics Analytics database at the Jackson Laboratory Genomic Medicine
"diseases of cellular proliferation" terms. (Susan Mockus)

Biomuta – HIVE project (cancer related variants):

Building a DO_Cancer_Slim (386 terms mapped to 187 term DO Cancer Slim)

Cancer terms from: COSMIC/UniProt/TCGA/IntOGen/ICGC/Publications

Collaborators: HIVE team, COSMIC, Disease Ontology, Novartis, EDRN

Project Team: Raja Mazumder, Tsung-Jung Wu, Krista Smith (GW University)

Coordination of Disease Concepts
HPO – MeSH – DO



Nucleic Acids Research

Nucleic Acids Research Advance Access published October 27, 2014

Nucleic Acids Research, 2014 **1**
doi: 10.1093/nar/gku1011

Disease Ontology 2015 update: an expanded and updated database of human diseases for linking biomedical knowledge through disease data

Warren A. Kibbe¹, Cesar Arze², Victor Felix², Elvira Mitraka², Evan Bolton³, Gang Fu³, Christopher J. Mungall⁴, Janos X. Binder^{5,6}, James Malone⁷, Drashti Vasant⁷, Helen Parkinson⁷ and Lynn M. Schriml^{2,8,*}



Gene Wiki



HPO
Human Phenotype
Ontology



Thank you

Download DO

<https://sourceforge.net/projects/diseaseontology/>

<https://github.com/obophenotype/human-disease-ontology>

[http://purl.obolibrary.org/obo/doid .owl](http://purl.obolibrary.org/obo/doid.owl)

DO Term Tracker - Submit new terms, definitions or suggestions for the Disease Ontology to the DO Term Tracker.

<http://sourceforge.net/p/diseaseontology/feature-requests/>