

# **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

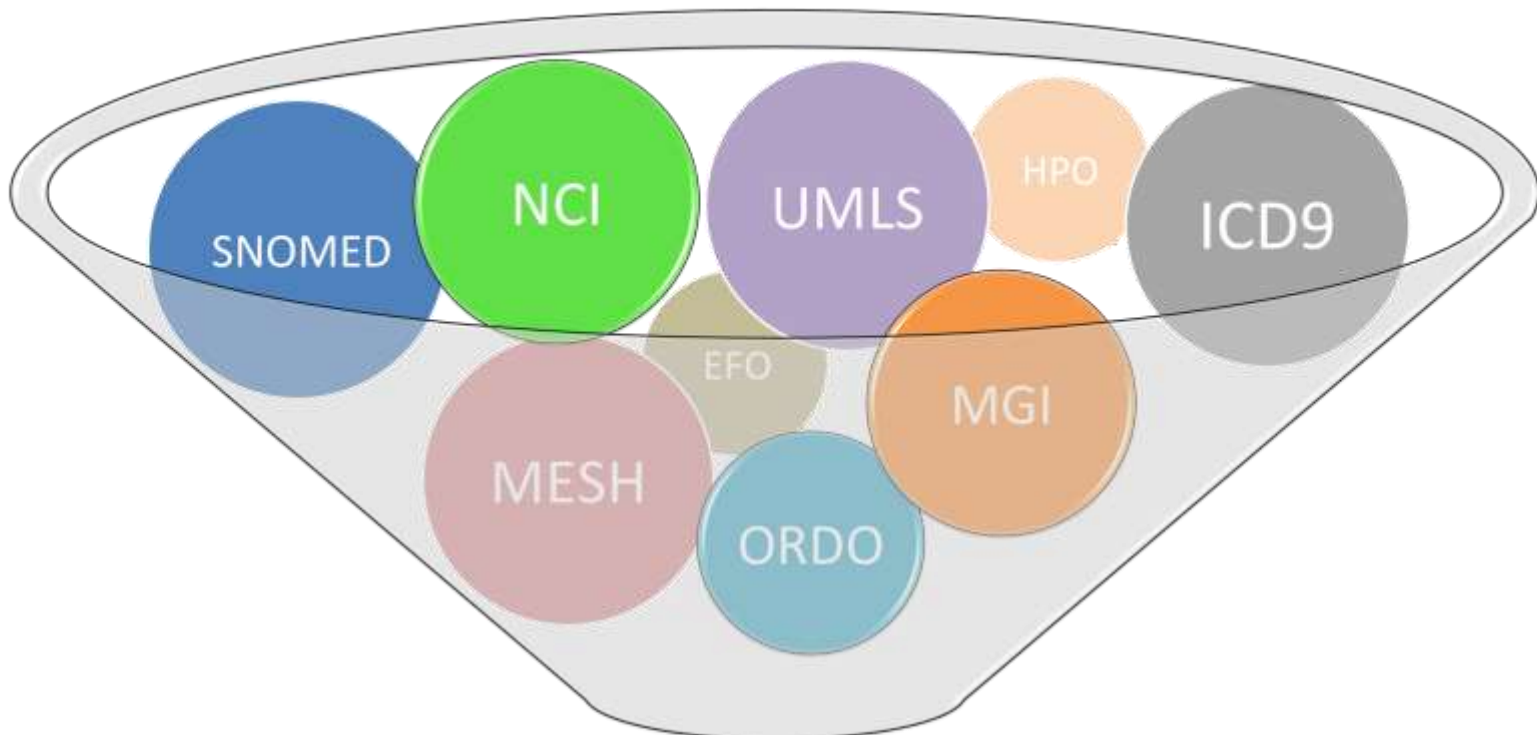
NCBI  
**NEW** MeSH  
NLM Controlled  
Vocabulary

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





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

DOI	DOI: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. <a href="http://www.cancer.gov/cancertopics/types/adrenocortical">http://www.cancer.gov/cancertopics/types/adrenocortical</a>
Xrefs	<a href="#">MSH:D018268</a> <a href="#">NCI:C9325</a> <a href="#">OMIM:202300</a> 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 <a href="#">adrenal cortex cancer</a>

Add an item to the term tracker

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	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<sup>KO17</sup>,

gene: Neuroligin 2

FBgn0031866



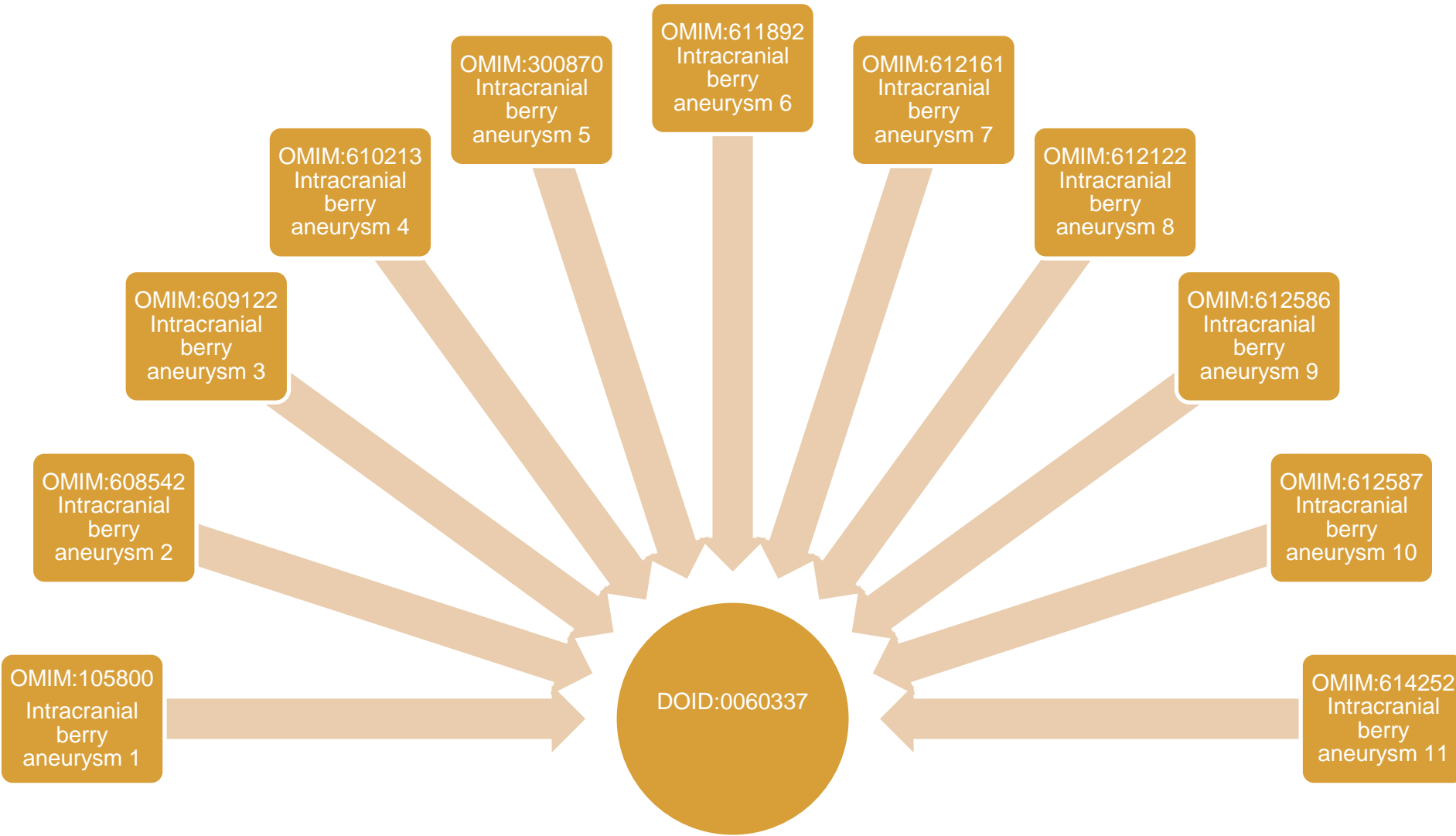
32 mouse models

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. Kibbe<sup>1</sup>, Cesar Arze<sup>2</sup>, Victor Felix<sup>2</sup>, Elvira Mitra<sup>2</sup>, Evan Bolton<sup>3</sup>, Gang Fu<sup>3</sup>, Christopher J. Mungall<sup>4</sup>, Janos X. Binder<sup>5,6</sup>, James Malone<sup>7</sup>, Drashti Vasant<sup>7</sup>, Helen Parkinson<sup>7</sup> and Lynn M. Schriml<sup>2,8,\*</sup>



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/>