

TACACAATCAGTTAGTTTCCACCGACAGTCCGAGAAACCATTCGACGGC  
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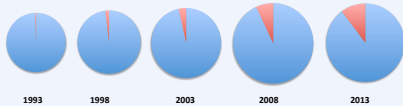


# FlyBase

# Human-health-related models in *Drosophila melanogaster*

## Introduction

The number and proportion of *Drosophila* articles published each year that mention disease has risen steadily. Many of these papers describe *Drosophila* models of human diseases or investigate functional conservation between human-fly orthologs. To help researchers find genes and alleles that model human diseases in *Drosophila* FlyBase has started to collect and display this information from primary research articles.



Papers containing "disease" in the abstract or title

## Human-health-related models in FlyBase

### What disease models are captured by FlyBase?

In our initial efforts to assemble data on disease models on *Drosophila* we have limited the data curation to **genetic models** which result in a **phenotype** that **recapitulates** some aspect of **human disease**. The genetic models we have applied this to are at the **allele** level i.e. at-locus-mutations and transgenes.

### What alleles are included?

- Disease-phenotype-causing- alleles with links to human disease.
- Alleles which modify the disease-model phenotype.

### How are alleles linked with disease?

- Alleles are associated with standardized human disease terms from the Disease Ontology (disease-ontology.org).
- An allele's relationship with a disease can be described as
  - a **model of**
  - a modifier: **ameliorates** or **exacerbates** a disease **modeled by** another allele

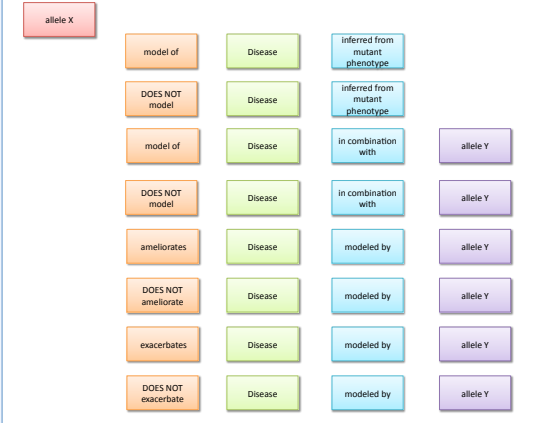
•When >1 allele models the disease, "**in combination with**" is used to describe their relationship.

•a **DOES NOT** prefix is used for cases when a particular allele fails to manifest the expected phenotype.

•Therefore using simple expressions there are multiple ways in which the relationship between an allele and disease can be described.



### Eight ways to describe the relationship between an allele and a human disease



### Finding disease-related papers

- To identify potential papers for disease curation we have:
  - Searched the archives for papers with "disease" in the title or abstract,
  - Examined papers known to use human transgenes,
  - Sought author help: Flagging "Human Disease" on our "Fast Track Your Paper" tool.
- From this we have been able to associate ~1000 alleles of over 500 genes, spanning ~100 diseases (DO terms).

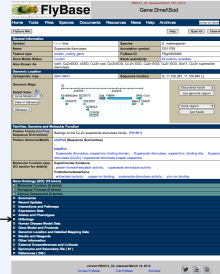


*Drosophila* has proven an incredibly tractable model for neurodegenerative disease. The fly has a complex brain and its short life-span enables the study of these diseases which often have an age-dependent pathology. It is therefore not surprising that the majority of *Drosophila* human-disease models relate to neurodegenerative diseases. This is illustrated in the word cloud above, generated from the Disease Ontology terms we have associated with alleles in FlyBase.

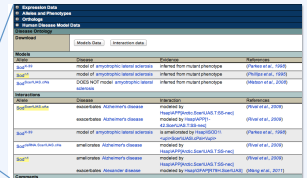
## How can I find human disease models in FlyBase?

### 1. Is a particular gene associated with a disease model?

In the **Gene Report** page for each gene there is now a **Human Disease Model Data** section.



The gene report page for the *D.mel* Sod gene. Clicking on a specific allele will take you to the associated **Allele Report** page.

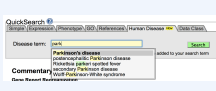


In the **Orthologs** section links can be found to **predicted human orthologs** and any related links to **OMIM**. Many disease models have been made by expressing human genes in *D.mel*, so don't forget to search for these genes too!

### 2. What genes/alleles are associated with a particular disease model?

There are two ways to directly search for a disease: **QuickSearch** and the dedicated Controlled Vocabulary search tool **TermLink**. Both have autocomplete functions, but **TermLink** will also allow you to explore the Disease Ontology hierarchy.

#### QuickSearch "Human Disease" tab



#### TermLink



### The disease term report page

Disease details: Name, Definition, Contact, Comment, etc.

Click for a list of all genes or alleles associated with this disease term

Disease Ontology hierarchy: The links here will take you to the associated disease term report page

Compound Statements: Open this section to find how the alleles are associated with the disease term. Clicking on the links will give you a list of these alleles.

Allele Hit list - a section of allele hit list for model of Parkinson's disease

Allele Report for park1

Phenotypic data: Look here for a description of the phenotype.

## This is just the beginning .....

- This newly compiled human disease data is in the FB2014\_02 release of FlyBase.
- Human disease curation is now an integral part of new paper curation at FlyBase.
- We are still looking at past papers to identify potential disease models and text-mining will be employed to help trawl the archives.
- A precompiled file will be available in the next release.
- Please help us make this resource as comprehensive and useful as possible by providing feedback, suggestions and alerting us to models we have missed. Use the **Contact FlyBase** link (found at the bottom of each page in FlyBase).



•Thank you to all authors who flagged "Human Disease" using the **Fast Track Your Paper** tool!

