Introduction
The number and proportion of Drosophila articles that mention disease is increasing year-on-year. 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.

Drosophila models of Human Disease in FlyBase
What disease models do we capture?
- Data on genetic models which result in a phenotype that recapitulates some aspect of human disease.
- The genetic models in this case are alleles.

How do we identify relevant papers?
- Newly published papers: authors tag the paper with “Human Disease” using our Fast Track Your Paper tool.
- Archive of previously published papers:
  - We searched for papers with “disease” in the title or abstract.
  - We examined papers known to use human transgenes.

What alleles are included?
- Drosophila genes: both at-locus mutations and transgenic constructs.
- Transgenic constructs expressing human genes.
- Alleles which cause a phenotype that models a human disease.
- Alleles which modify a disease-model phenotype.

How do we link alleles to disease?
- We associate the allele with standardized human disease terms from the Disease Ontology (http://disease-ontology.org).
  - We record whether the allele is:
    - A model of the human disease.
    - OR
    - A modifier, which ameliorates or exacerbates a disease modeled by another allele.
- When >1 allele models the disease, we record that they act “in combination with” each other.
- We use a DOES NOT preface to record unexpected negative results e.g. when a fly ortholog of a disease-causing human gene does not recapitulate the disease phenotype.
- Using these simple expressions there are multiple ways in which the relationship between an allele and a disease can be described.

The data so far
- FlyBase started capturing disease annotations at the beginning of 2014.
- The FB 2014.05 (September 9th 2014) release contains:
  - ~2,500 disease annotations from ~500 references.
  - ~1,500 alleles from ~750 genes are annotated as either a human-disease model or a modifier.
- Models of 126 different human diseases have been annotated.
- Approximately two-thirds of the models are of neurological diseases.

How to find human disease models in FlyBase
1. Is a particular gene/allele associated with a disease model?
   There is now a Human Disease Model Data section in each Gene and Allele Report.

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. Vocabularies. Both have autocompletion functions, but Vocabularies will also allow you to explore the Disease Ontology hierarchy.

3. How do I get the complete set of data?
   Download the precomputed file.

Future Improvements
- We are still looking at past papers to identify potential disease models and text-mining will be employed to help trawl the archives.
- Please help us make this resource as comprehensive and useful as possible by providing feedback, suggestions and alerting us to disease models we have missed. Use the Contact Flybase link (found at the bottom of each page in FlyBase).

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