# Finding Human Disease Models in FlyBase

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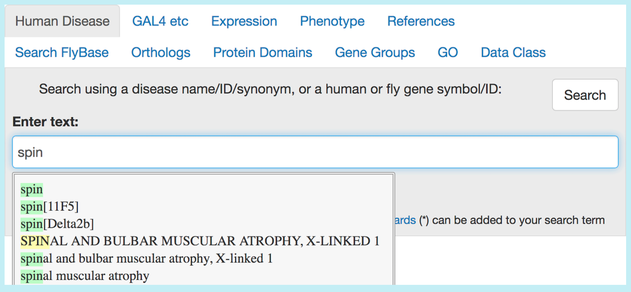
## The Human Disease QuickSearch Tab

FlyBase has updated the Human Disease QuickSearch tab to allow searches for Human Disease Model Reports, Disease Ontology terms, disease-associated genes, and alleles used to model disease.




The original **Human Disease** tab allowed only searches for Disease Ontology (DO) terms, using only DO terms or DOIDs. In the updated Human Disease tab, it is possible to search all FlyBase human disease model data using almost any disease-related search term, including disease synonyms; for example, the search string “Lou Gehrig Disease” will return amyotrophic lateral sclerosis.

**You can search by disease using:**
- Disease Ontology (DO) term or DOID
- Human Disease Model name or ID
- OMIM phenotype term or ID
- disease synonym

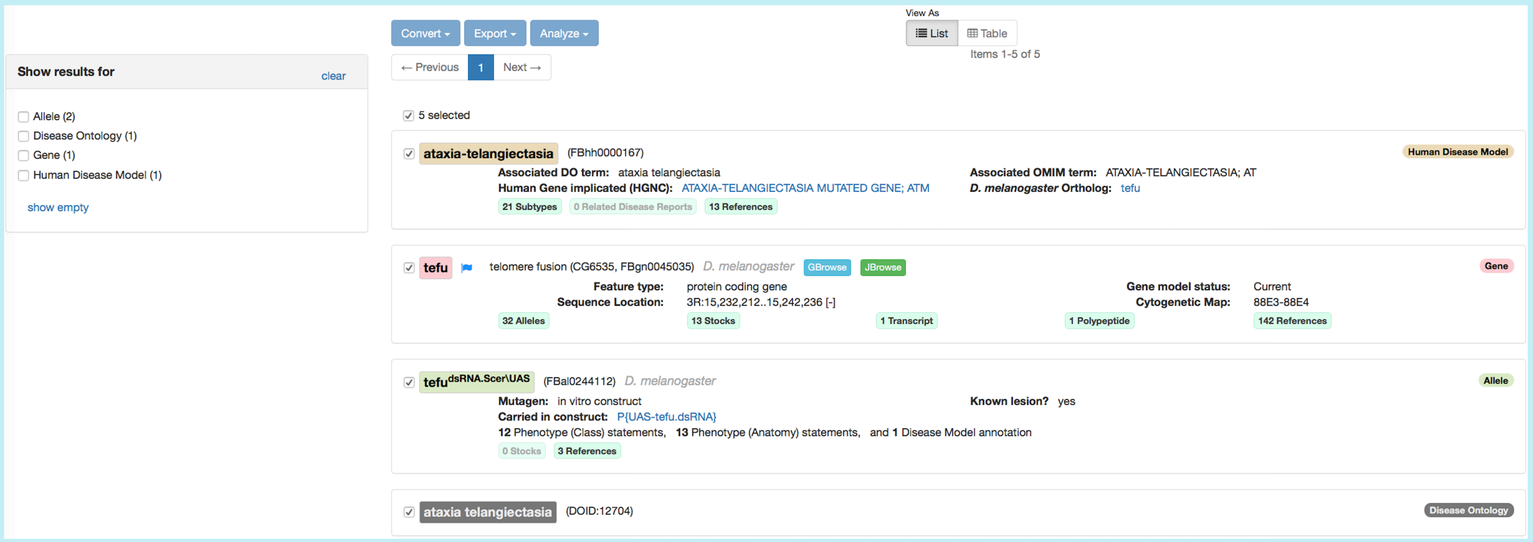
**You can search by human disease-associated gene using:**
- HGNC symbol or ID
- OMIM genotype symbol or ID

**You can search by *Drosophila melanogaster* gene using:**
- FlyBase gene symbol, name, or FBgn identifier.

Autocomplete for the **Human Disease** tab is exceptionally robust, and works for multiple input classes at once; as shown in the example above, the string “spin” simultaneously triggers autocompletion of gene symbols, allele symbols, OMIM diseases terms, DO terms, and Human Disease Model names.

**Autocomplete works for:**
- Disease Ontology terms and DOIDs
- Human Disease Model names and IDs
- OMIM phenotype and genotype terms and IDs
- HGNC gene symbols and IDs
- FlyBase gene symbols, names, and IDs
- FlyBase allele symbols and IDs

## Human Disease Hit-lists: Now With More Data Classes




The Human Disease hit-list now features four classes of data: Human Disease Models, Disease Ontology terms, genes, and alleles. This hit-list resulted from the search “ataxia telangiectasia”, for which the hits are the Human Disease Model ataxia-telangiectasia, the *Drosophila melanogaster* gene tefu associated with that model, the DO term ataxia telangiectasia, and the allele tefu$^{dsRNA.Scer\UAS}$, which is annotated with that DO term.

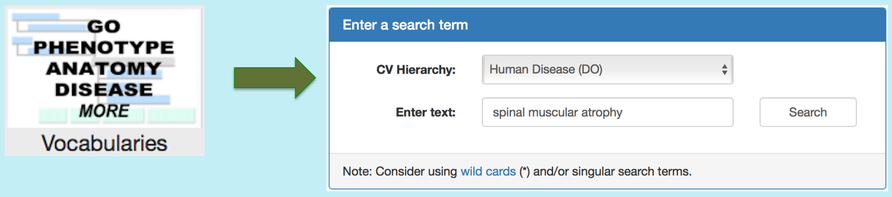
Like all FlyBase faceted hit-lists, a user can choose to display a subset of data classes. Filtering the hit-list to a single data class allows you to further convert the results to another data class, export to a file or to another FlyBase tool, analyze the results in a data class specific manner, or display the hit-list as a table.

**Human Disease Model Results**

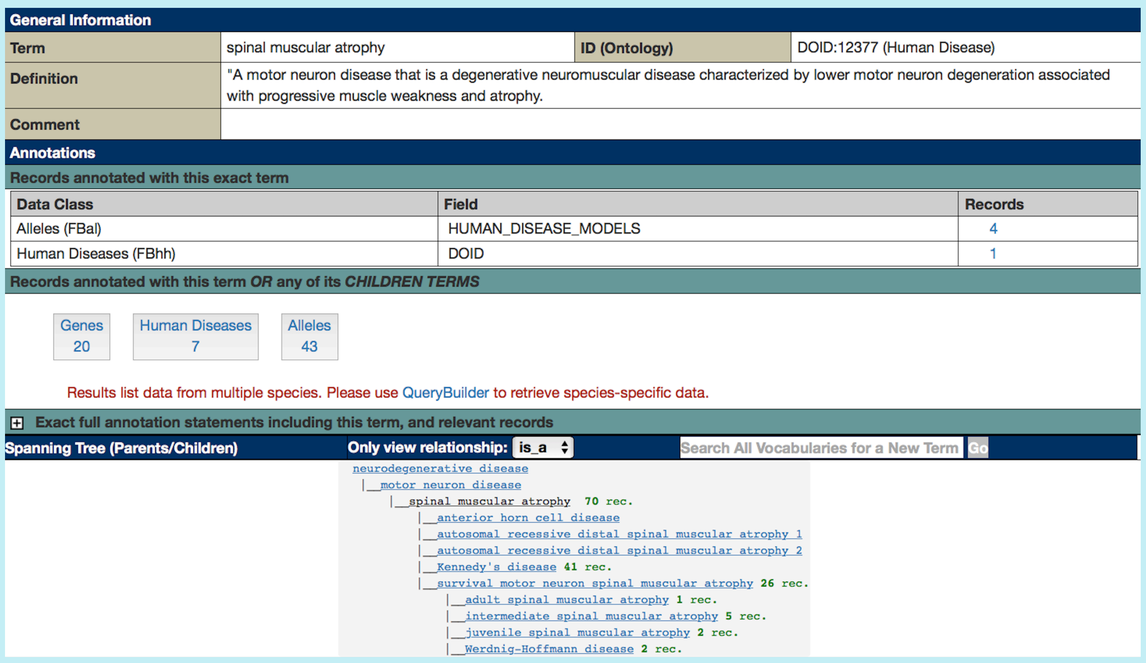
| Name | DO Term | OMIM ID | *D. melanogaster* gene | Human Genes Implicated (HGNC) | # Subtypes | # of Related Disease Terms | # Refs |
|---|---|---|---|---|---|---|---|
| amyotrophic lateral sclerosis 1 | amyotrophic lateral sclerosis type 1 | 105400 | Sod1 | SUPEROXIDE DISMUTASE 1; SOD1 | 24 | 0 | 36 |
| amyotrophic lateral sclerosis 8 | amyotrophic lateral sclerosis type 8 | 608627 | Vap33 | VESICLE-ASSOCIATED MEMBRANE PROTEIN-ASSOCIATED PROTEIN B; VAPB | 24 | 1 | 22 |
| amyotrophic lateral sclerosis 17 | amyotrophic lateral sclerosis type 17 | 614696 | | CHMP FAMILY, MEMBER 2B; CHMP2B | 24 | 1 | 6 |
| amyotrophic lateral sclerosis 6 | amyotrophic lateral sclerosis type 6 | 608030 | caz | FUSED IN SARCOMA; FUS | 24 | 2 | 51 |
| amyotrophic lateral sclerosis 18 | amyotrophic lateral sclerosis type 18 | 614808 | chic | PROFILIN 1; PFN1 | 24 | 1 | 7 |
| amyotrophic lateral sclerosis 2 | amyotrophic lateral sclerosis type 2 | 205100 | Als2 | ALSIN | 24 | 0 | 5 |

This hit-list, resulting from the search “amyotrophic lateral sclerosis”, has been filtered to display only Human Disease Models. The table view for this data class highlights the relationships between Human Disease Models, DO terms, and OMIM phenotypes, and displays the associated human disease gene and its orthologous *Drosophila melanogaster* gene.

## Using the Vocabularies Tool to Find Disease Model Information

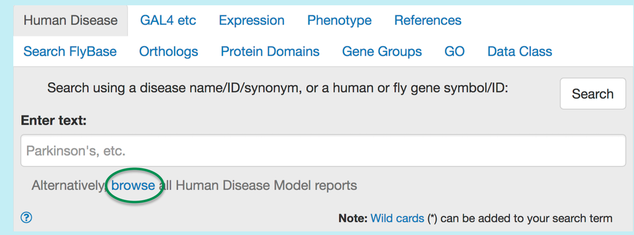



The **Vocabularies** tool can be accessed using the button on the FlyBase home page, or from the Tools drop-down menu in the FlyBase toolbar. Select the **Human Disease (DO)** CV Hierarchy and enter a search term; autocomplete is enabled for DO terms.

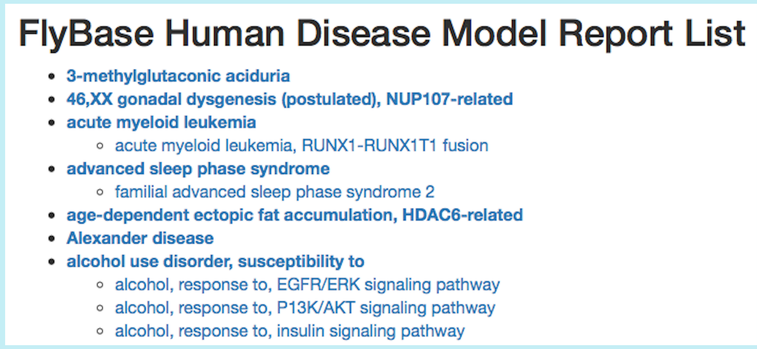



The Disease Ontology term report provides further access to disease model data. The spanning tree allows the user to browse the DO hierarchy, which displays both less specific parental disease categories, and more specific child disease terms. The buttons above the spanning tree lead to hit-lists of genes associated with, Human Disease Model Reports linked to, and alleles annotated with the DO term or its children.

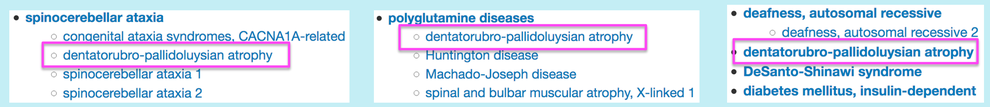
## The Human Disease Model Report Index




The **Human Disease Model Report List** is accessed via a link on the **Human Disease** QuickSearch tab.




The **Human Disease Model Report List** consists of links to those reports. Many disease model reports are redundantly listed, so that this resource also serves as an index, allowing a user to browse to a disease from multiple points. Diseases may be listed as a specific subtype of a disease, by mechanistic cause, by symptomatic group, or as part of a major disease classification.




In the example above, **dentatorubro-pallidoluysian atrophy** is a subtype of **spinocerebellar ataxia**, is one of several **polyglutamine diseases**, and is listed alphabetically by name.

Major disease classifications under which a user might find a disease model of interest include epilepsy, cancer, cardiac dysfunction, kidney disease, and muscular dystrophy, among others.