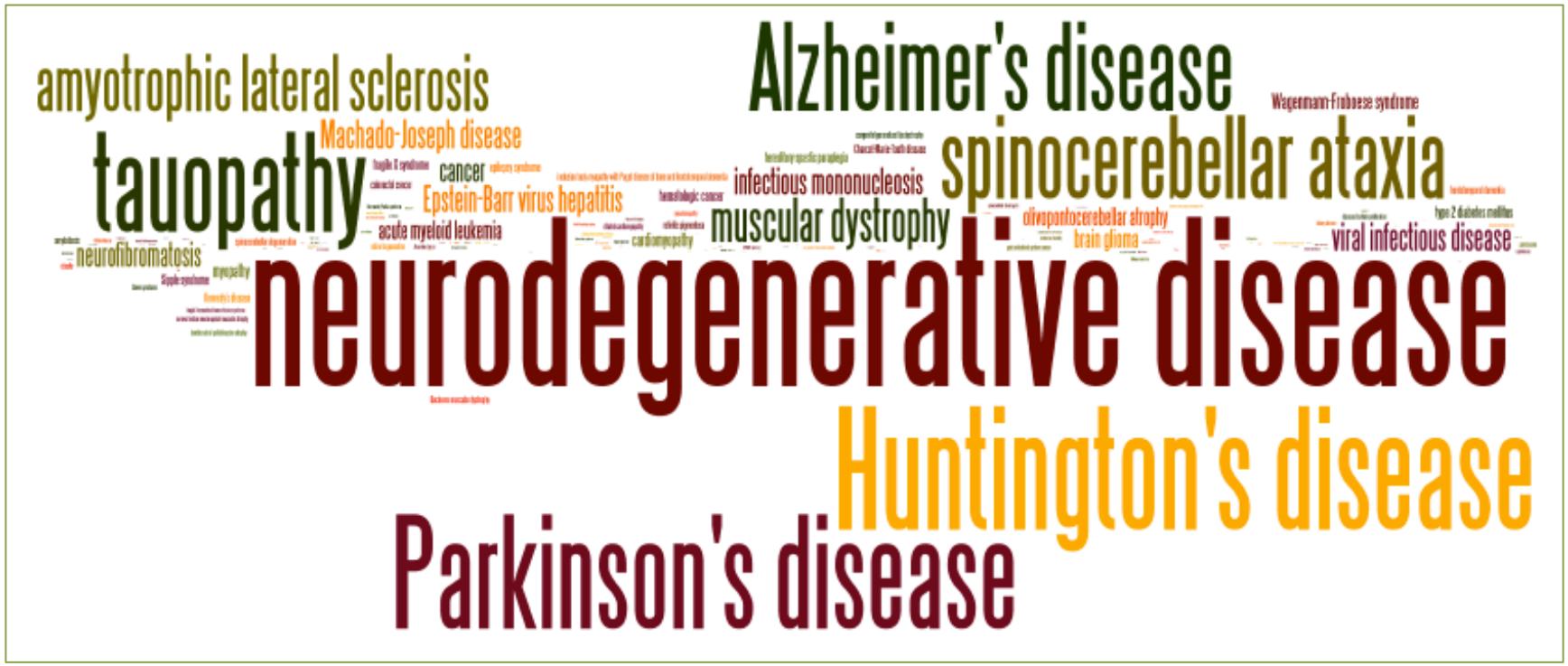
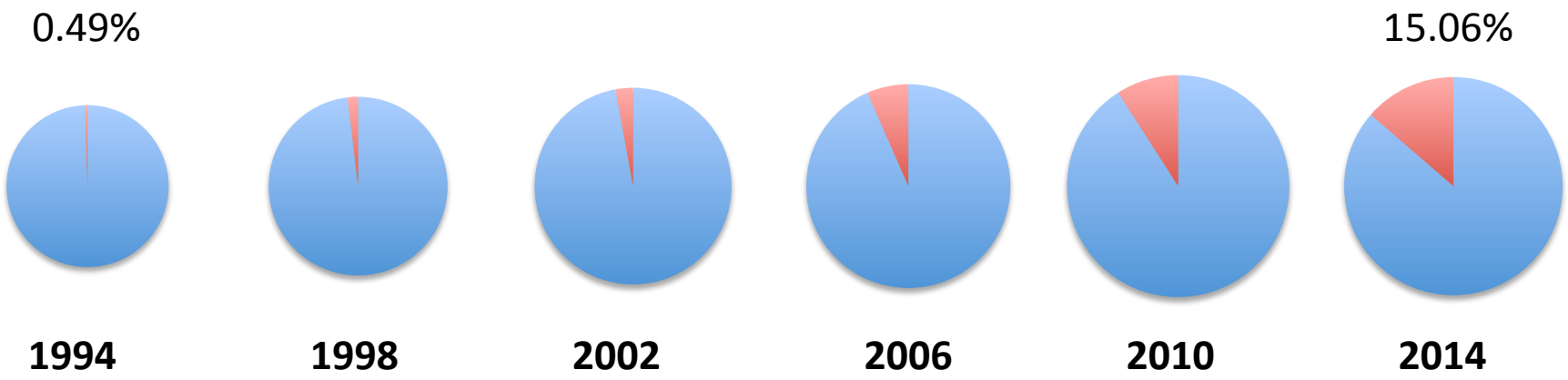
Human Disease models and Gene Groups

Human Disease models



Human Disease models



- *Drosophila* papers containing “disease” in the abstract or title

What disease models do we capture ?

- Data on **genetic models** which result in a **phenotype** that **recapitulates** some aspect of **human disease**.

- At the **allele** level
  - Classical and transgenic constructs

How do we identify relevant papers ?

Newly published papers

Authors tag the paper with "Human Disease" using our **Fast Track Your Paper** tool

Text-mining non-author curated papers for human disease

Human disease relevant papers

Archive of previously published papers

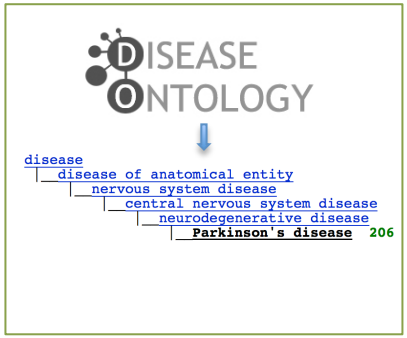
We searched for papers with "disease" in the title or abstract.

We examined papers known to use human transgenes.

Text-mining non-author curated papers for human disease

How do we link alleles to disease ?

We associate the allele with standardized human disease terms from the Disease Ontology



Bsk[1] allele report

Disease Ontology		
Models		
Disease	Evidence	References
model of Parkinson's disease	inferred from mutant phenotype	(*Inamdar et al., 2014*)
Interactions		
Disease	Interaction	References
ameliorates Alzheimer's disease	modeled by Hsap\APP$^{1\text{-}42.Scer\UAS.T:SS\text{-}nec}$	(*Hong et al., 2012*)
ameliorates retinitis pigmentosa	modeled by ninaE^{P37H}, ninaE^{17}	(*Griciuc et al., 2014*)
Comments		

DO maintained in collaboration with Lynn Schriml

How do we link alleles to disease ?

salr^{FCK-25} allele report

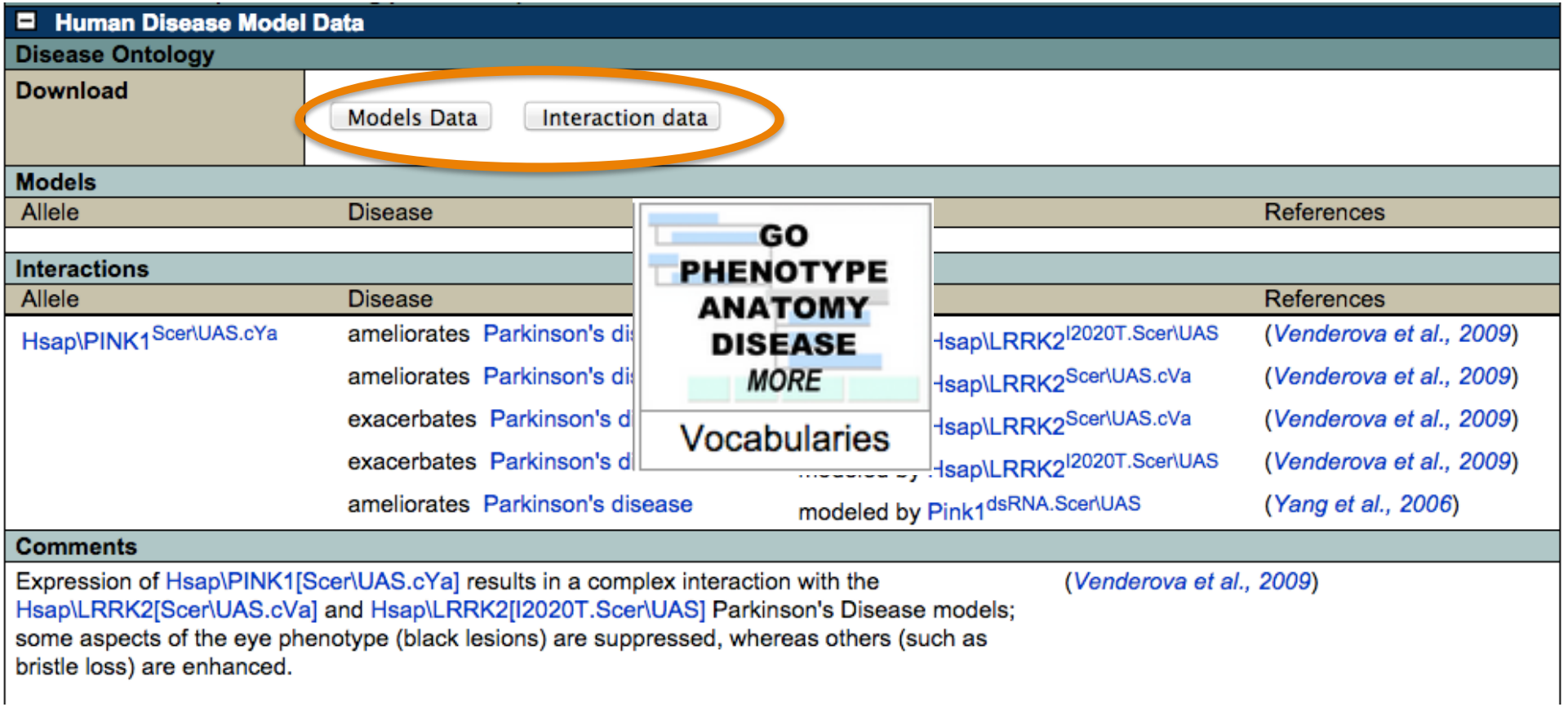
Human Disease Model Data		
Disease Ontology		
Models		
Disease	Evidence	References
model of Townes-Brocks syndrome	in combination with salm^{FCK-25}	(*Si-Dong et al., 2003*)
Interactions		
Disease	Interaction	References
Comments		

Hsap/APP$^{F20E.Scer\UAS.T:SS-nec}$ allele report

Disease Ontology		
Models		
Disease	Evidence	References
DOES NOT model Alzheimer's disease	inferred from mutant phenotype	(*Luheshi et al., 2007*)

Display and searching

Pink1 gene report



Relationships	
Is a	synucleinopathy
Part of	

Synonyms & Secondary IDs	
Synonyms	
	paralysis agitans" EXACT [CSP2005:2057-3689] Parkinson disease" EXACT []
Secondary IDs	

External Crossreferences & Linkouts	
	EFO:0002508 ICD9CM:332 ICD9CM:332.0 KEGG:05012 MSH:D010300 NCI:C26845 OMIM:168600 OMIM:168601 OMIM:260300 OMIM:556500 OMIM:600116 OMIM:605543 OMIM:605909 OMIM:606324 OMIM:607060 OMIM:607688 OMIM:610297 OMIM:612953 OMIM:613643 SNOMEDCT_2010_1_31:154999006 SNOMEDCT_2010_1_31:155000006 SNOMEDCT_2010_1_31:155002003 SNOMEDCT_2010_1_31:192825001 SNOMEDCT_2010_1_31:192831003 SNOMEDCT_2010_1_31:49049000 UMLS_CUI:C0030567
Linkouts	

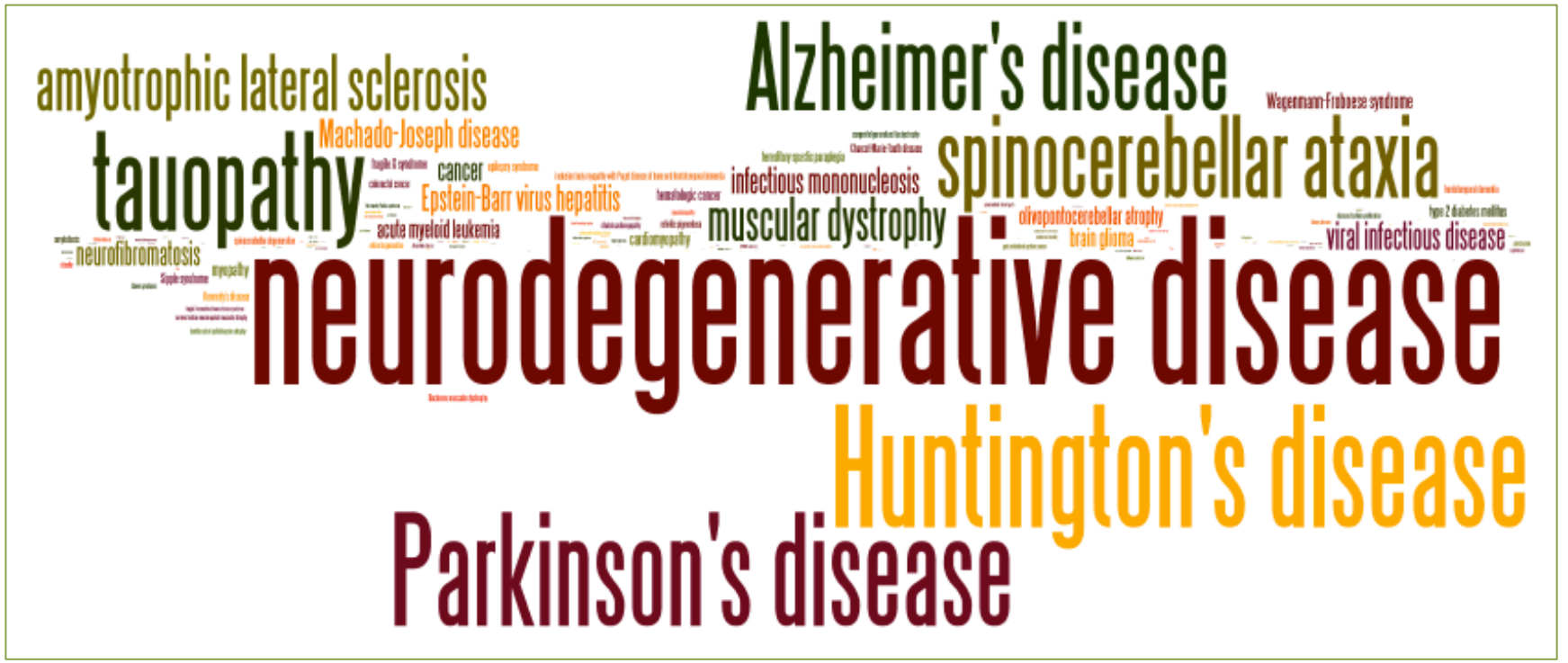
Annotation effort so far

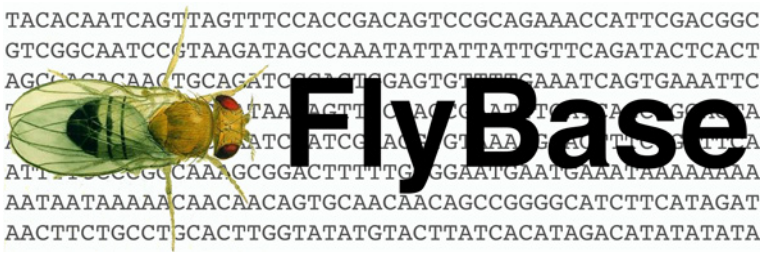
FlyBase started capturing disease annotations at the beginning of 2014.

The FB_2015_01 (February 24th 2015) release contains:

- ~**2,800** disease annotations from ~**600** references.
- **>1,900** alleles from **876** genes are annotated as either a human-disease model or a modifier. Models of **151** different human diseases have been annotated.

- Approximately **two-thirds** of the models are of neurological diseases.


amyotrophic lateral sclerosis
Alzheimer's disease
Wagenmann-Frobiese syndrome
Machado-Joseph disease
tauopathy
spinocerebellar ataxia
cancer
Epstein-Barr virus hepatitis
infectious mononucleosis
muscular dystrophy
olivopontocerebellar atrophy
brain glioma
viral infectious disease
type 2 diabetes mellitus
acute myeloid leukemia
neurofibromatosis
neurodegenerative disease
Huntington's disease
Parkinson's disease



Gene Groups in FlyBase

Steven Marygold
Helen Attrill
Kathleen Falls
Josh Goodman
Gillian Millburn
FlyBase consortium

What is a ‘Gene Group’?

Genes or gene products that are acknowledged to form a biological group

i. Gene families (actins, olfactory receptors…)
ii. Protein complex subunits (proteasome, ribosome…)
iii. Other functional grouping (ubiquitin E3 ligases, cadherins…)

- Described in research/review article(s)
- *D. melanogaster* only

RESEARCH ARTICLES

Molecular Evolution and Functional Diversification of Fatty Acid Desaturases after Recurrent Gene Duplication in *Drosophila*

Shu Fang,[2] Chau-Ti Ting,†‡[2] Cheng-Ruei Lee,* Kuang-Hsi Chu,‡ Chuan-Chan Wang,*[1] and Shun-Chern Tsaur**

RESEARCH ARTICLE

The four aldehyde oxidases of *Drosophila melanogaster* have different gene expression patterns and enzyme substrate specificities

The Molecular Evolution of Cytochrome P450 Genes within and between *Drosophila* Species

Robert T. Good†, Lydia Gramzow†,[1], Paul Battlay, Tamar Sztal[2], Philip Batterham, and Charles Robin*

Characterization of the adenosine deaminase-related growth factor (ADGF) gene family in Drosophila

Stephanie A. Maier, Lynn Podemski, Sean W. Graham, Heather E. McDermid, John Locke*

Review

Drosophila metalloproteases in development and differentiation: The role of ADAM proteins and their relatives

Heiko Meyer, Mareike Panz, Stefanie Albrecht, Maik Drechsler, Shuoshuo Wang, Mirko Hüsken, Christine Lehmacher, Achim Paululat*

Developmental and functional studies of the SLC12 gene family members from *Drosophila melanogaster*

Qifei Sun,[1] E. Tian,[2] R. James Turner,[1] and Kelly G. Ten Hagen[2]

An Inventory of Peroxisomal Proteins and Pathways in *Drosophila melanogaster*

Joseph E. Faust, Avani Verma, Chengwei Peng and James A. McNew*

three main routes. Most proteins are targeted to isomes via a peroxisomal targeting signal type 1 at their carboxy-termini that matches SKL or a con

Characterization of Maltase Clusters in the Genus *Drosophila*

Marek Gabriško · Štefan Janeček

Receptor Tyrosine Kinases in *Drosophila* Development

Richelle Sopko[1] and Norbert Perrimon[1,2]

Functional Characterization and Expression Analysis of Members of the UDP-GalNAc:Polypeptide *N*-Acetylgalactosaminyltransferase Family from *Drosophila melanogaster**

Received for publication, April 11, 2003, and in revised form, June 20, 2003
Published, JBC Papers in Press, June 26, 2003, DOI 10.1074/jbc.M303836200

Kelly G. Ten Hagen‡§, Duy T. Tran‡, Thomas A. Gerken¶, David S. Stein‖, and Zhenyu Zhang‖

Conservation of the Protein Composition and Electron Microscopy Structure of *Drosophila melanogaster* and Human Spliceosomal Complexes▽†

Nadine Herold,[1] Cindy L. Will,[1] Elmar Wolf,[1] Berthold Kastner,[1] Henning Urlaub,[2] and Reinhard Lührmann[1]*

Neprilysins: An Evolutionarily Conserved Family of Metalloproteases That Play Important Roles in Reproduction in *Drosophila*

Jessica L. Sitnik,* Carmen Francis,†,‡,[1] Korneel Hens,†,‡ Roger Huybrechts,§ Mariana F. Wolfner,*,[3] and Patrick Callaerts†,‡,[3]

Functional studies of *Drosophila* zinc transporters reveal the mechanism for dietary zinc absorption and regulation

Qiuhong Qin, Xiaoxi Wang and Bing Zhou*

The Genetic Architecture of Degenerin/Epithelial Sodium Channels in *Drosophila*

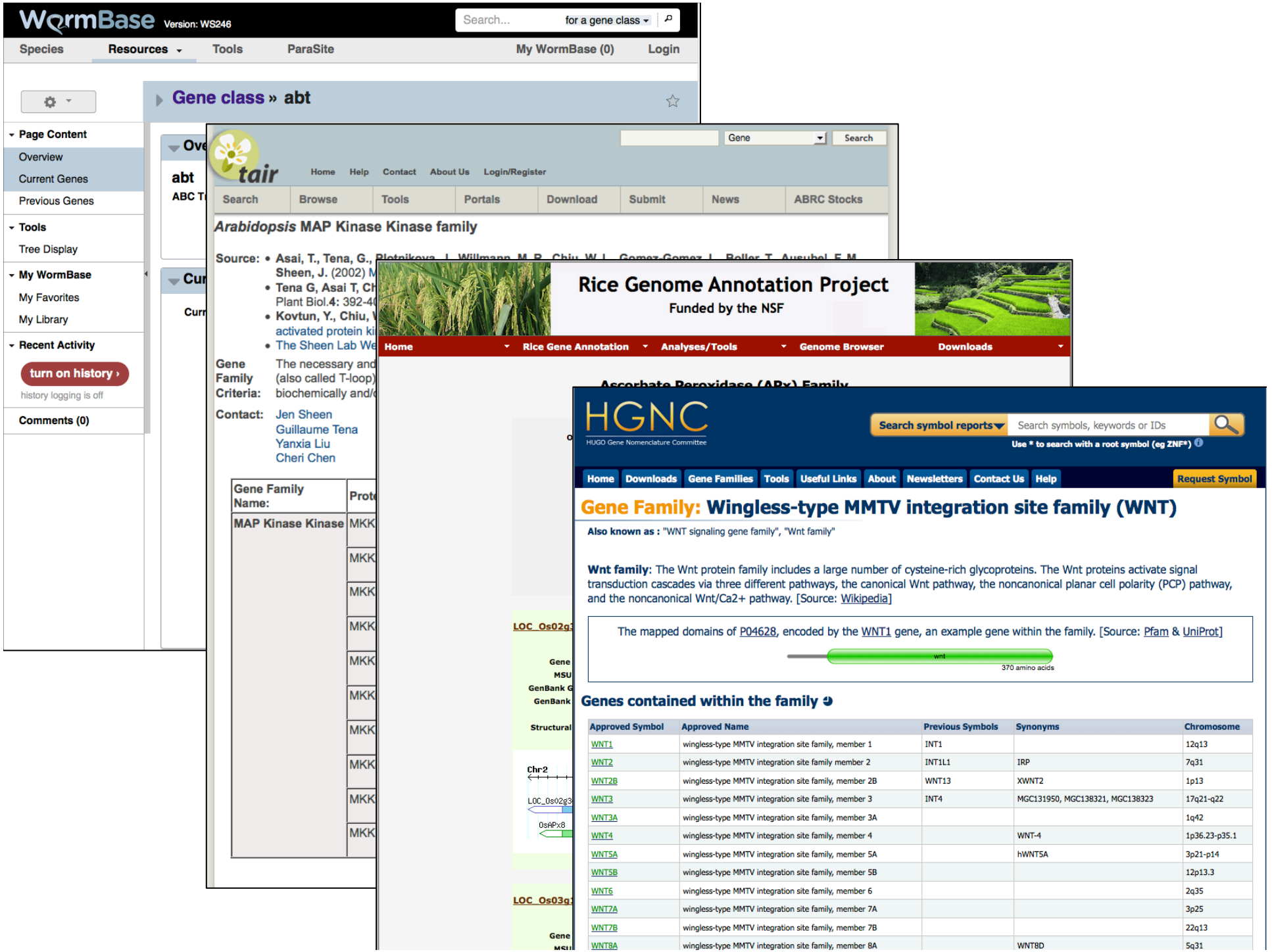
Kathleen M. Zelle,[1] Beika Lu,[1,2] Sarah C. Pyfrom, and Yehuda Ben-Shahar[3]

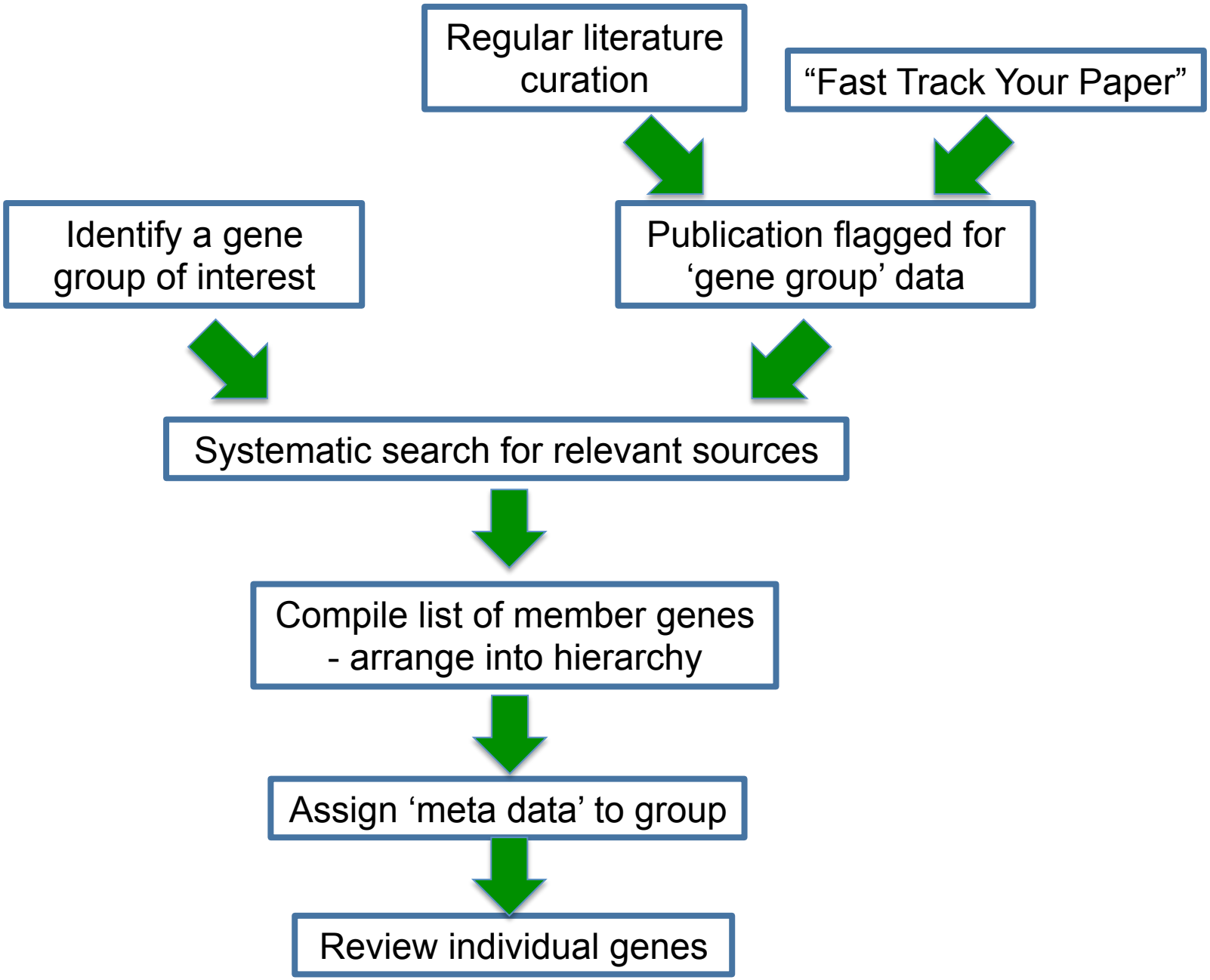
Example: Myosin light chains

FB Symbol	FB Name	Shared Protein domains	Shared GO annotations
Mlc-c	Myosin light chain cytoplasmic	• EF-hand domain • EF-hand domain pair	• myosin complex • calcium ion binding
Mlc1	Myosin alkali light chain 1		
Mlc2	Myosin light chain 2		
sqh	spaghetti squash		

How obtain this list?

1. Gene symbol/name/synonym?
2. Protein domain(s)?
3. GO annotations?
4. Find relevant publication in FlyBase -> associated genes?


WormBase Version: WS246
Search... for a gene class
Species Resources Tools ParaSite My WormBase (0) Login
Gene class » abt
Page Content
Overview
Current Genes
Previous Genes
Tools
Tree Display
My WormBase
My Favorites
My Library
Recent Activity
turn on history ›
history logging is off
Comments (0)
tair
Home Help Contact About Us Login/Register
Search Browse Tools Portals Download Submit News ABRC Stocks
Arabidopsis MAP Kinase Kinase family
Source:
Gene Family Criteria:
Contact: Jen Sheen
Guillaume Tena
Yanxia Liu
Cheri Chen
Gene Family Name:
MAP Kinase Kinase
Rice Genome Annotation Project
Funded by the NSF
Home Rice Gene Annotation Analyses/Tools Genome Browser Downloads
HGNC
HUGO Gene Nomenclature Committee
Search symbol reports
Search symbols, keywords or IDs
Use * to search with a root symbol (eg ZNF*)
Home Downloads Gene Families Tools Useful Links About Newsletters Contact Us Help
Request Symbol
Gene Family: Wingless-type MMTV integration site family (WNT)
Also known as : "WNT signaling gene family", "Wnt family"
Wnt family: The Wnt protein family includes a large number of cysteine-rich glycoproteins. The Wnt proteins activate signal transduction cascades via three different pathways, the canonical Wnt pathway, the noncanonical planar cell polarity (PCP) pathway, and the noncanonical Wnt/Ca2+ pathway. [Source: Wikipedia]
The mapped domains of P04628, encoded by the WNT1 gene, an example gene within the family. [Source: Pfam & UniProt]
wnt
370 amino acids
Genes contained within the family
Approved Symbol | Approved Name | Previous Symbols | Synonyms | Chromosome
WNT1 | wingless-type MMTV integration site family, member 1 | INT1 | | 12q13
WNT2 | wingless-type MMTV integration site family member 2 | INT1L1 | IRP | 7q31
WNT2B | wingless-type MMTV integration site family, member 2B | WNT13 | XWNT2 | 1p13
WNT3 | wingless-type MMTV integration site family, member 3 | INT4 | MGC131950, MGC138321, MGC138323 | 17q21-q22
WNT3A | wingless-type MMTV integration site family, member 3A | | | 1q42
WNT4 | wingless-type MMTV integration site family, member 4 | | WNT-4 | 1p36.23-p35.1
WNT5A | wingless-type MMTV integration site family, member 5A | | hWNT5A | 3p21-p14
WNT5B | wingless-type MMTV integration site family, member 5B | | | 12p13.3
WNT6 | wingless-type MMTV integration site family, member 6 | | | 2q35
WNT7A | wingless-type MMTV integration site family, member 7A | | | 3p25
WNT7B | wingless-type MMTV integration site family, member 7B | | | 22q13
WNT8A | wingless-type MMTV integration site family, member 8A | | WNT8D | 5q31


Regular literature curation
“Fast Track Your Paper”
Identify a gene group of interest
Publication flagged for ‘gene group’ data
Systematic search for relevant sources
Compile list of member genes - arrange into hierarchy
Assign ‘meta data’ to group
Review individual genes

Gene Group Report: CLASS B GPCRs, SUBFAMILY B1

General Information

Name	CLASS B GPCRs, SUBFAMILY B1	**Species**	*D. melanogaster*
Symbol	GPCR-B1	**FlyBase ID**	FBgg0000046
Date last reviewed	2013-08-15	**Number of members**	5

Description

Description	The B1 subfamily of Class B GPCRs are largely classical hormone receptors. (Adapted from FBrf0147071 and FBrf0221117).
Notes on membership	
Source Material	The CLASS B GPCRs, SUBFAMILY B1 Gene Group has been compiled by FlyBase curators using the membership given in the following publication(s): *Hauser et al., 2006*, *Harmar, 2001*, *Li et al., 2013*, and *Hauser et al., 2006*.

Key Gene Ontology (GO) terms

Molecular Function	protein-hormone receptor activity
Biological Process	G-protein coupled receptor signaling pathway
Cellular Component	integral component of plasma membrane

Related Gene Groups

Parent group(s)	CLASS B GPCRs

Members (5)

Export Genes to: Hitlist | Batch Download

Gene Symbol	Gene Name	Annotation ID	Source Material
Dh31-R	Diuretic hormone 31 Receptor	CG32843	(*Hauser et al., 2006*, *Harmar, 2001*, *Li et al., 2013*, *Hauser et al., 2006*)
Dh44-R1	Diuretic hormone 44 receptor 1	CG8422	(*Hauser et al., 2006*, *Harmar, 2001*, *Li et al., 2013*, *Hauser et al., 2006*)
Dh44-R2	Diuretic hormone 44 receptor 2	CG12370	(*Hauser et al., 2006*, *Harmar, 2001*, *Li et al., 2013*, *Hauser et al., 2006*)
hec	hector	CG4395	(*Hauser et al., 2006*, *Harmar, 2001*, *Li et al., 2013*, *Hauser et al., 2006*)
Pdfr	Pigment-dispersing factor receptor	CG13758	(*Hauser et al., 2006*, *Harmar, 2001*, *Li et al., 2013*, *Hauser et al., 2006*)

Recent Updates

External Data

Orthologous Group(s)	Human Class B GPCRs
Other resources(s)	

Synonyms and Secondary IDs

References (5)

‘Group curation’ - Added benefits

1. Gene Ontology (GO) annotation review
2. Nomenclature review
3. Un-annotated genes review

Summary

Availability:

- Human disease model data available now
- Gene Group Reports planned for April 2015

Feedback:

- Flag data types using ‘Fast Track Your Paper’
- Use ‘Contact FlyBase’ link
- Join ‘FlyBase Community Advisory Group’

National Human Genome Research Institute

MRC Medical Research Council