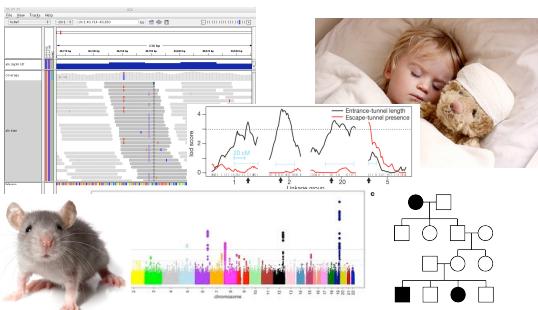




The Human-Mouse: Disease Connection in MGI (BETA)

Short Course Bioinformatics Workshops
July 2014

Connecting Mouse Models to Human Disease



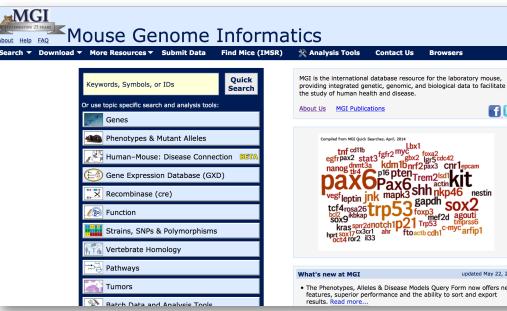
A detailed figure showing a pedigree chart with symbols for affected (filled) and unaffected (open) individuals. Below it is a histogram of chromosomes (1-22, X, Y) with a color scale at the bottom. To the right is a graph with 'Entrance-tunnel length' on the y-axis (0-4) and 'Entrance-tunnel presence' on the x-axis (0-20). A child in bed with a teddy bear is shown in the background.

HMDc in a Nutshell



- Search for mouse models by
 - gene symbol (mouse or human)
 - exome results (VCF file)
 - genome locations
 - key words
- Quick access to phenotype and disease annotations, references, mouse model availability

Mouse Genome Informatics



MGI is the international database resource for the laboratory mouse, providing integrated genetic, genomic, and biological data to facilitate the study of human health and disease.

<http://www.informatics.jax.org>

Human-Mouse: Disease Connection

Relating human diseases and mouse models

Search by genes

Search by genome locations

Search by disease or phenotype terms

Upload a VCF File

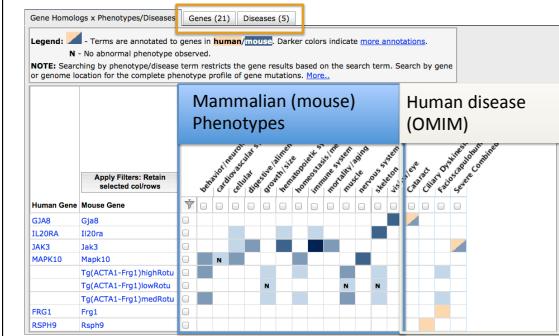
Spotlight on mouse models of human disease

Human T-Cell Immunodeficiency, Congenital Alopecia, and Nail dystrophy (OMIM: 601703)

Introduction to Mouse Genetics

Glossary of Terms

View associated gene, phenotype and disease results



Search by Gene Symbol

Search by genes

Ex: Gja8, Mapk10, Rspn9, Il2ra, Jak3, Frg1

Search by genome locations

Ex: Chr12:30000000-100000000

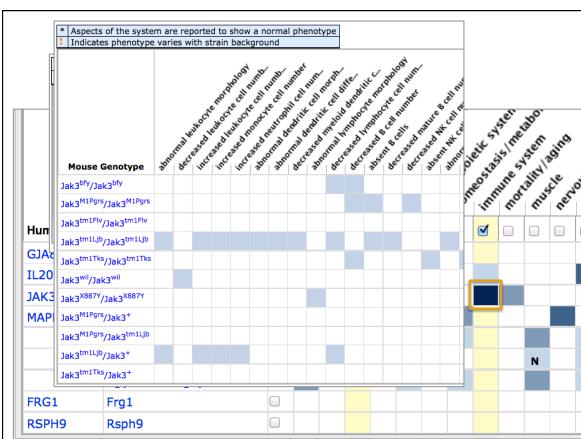
Need to convert genome build? Use the [converter tool](#).

Search by disease or phenotype terms

Ex: diabetes, 105830

Select from autocomplete or continue typing. Use quotes for exact match.

GO, Reset, BETA



Disease associations

Data for GJA8, Gja8 and Cataract

Mouse Genotype	Cataract, Multiple Types, C-
Gja8 ^{No2} /Gja8 ^{No2}	
Gja8 ^{R205G} /Gja8 ^{R205G}	
Gja8 ^{tm1Paul} /Gja8 ^{tm1Paul}	
Gja8 ^{No2} /Gja8 ⁺	
Human Gene	
GJA8	

Gene Homologs x Phenotypes/Diseases [Genes (21) | Diseases (5)]

Legend: N - Term is annotated to genes in human/mouse. Darker colors indicate more annotations.

NOTE: Searching by phenotype/disease term restricts the gene results based on the search term. Search by gene or genomic location for the complete phenotype profile of gene mutations. [More...](#)

Apply Filters: Retain selected columns

Human Gene	Mouse Gene	Behavior/mentality	Cardiovascular system	Digestive system	Endocrine system	Genitourinary system	Immune system	Integumentary system	Musculoskeletal system	Neurological system	Respiratory system	Stomach	Urinary system	Cataract	Ciliary Dyskinesia, Primary	Fascioscapulohumeral Muscular Dystrophy	Severe Combined Immunodeficiency
GJA8	Gja8	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N
IL20RA	I20ra																
JAK3	Jak3																
MAPK10	Mapk10	Tg(ACTA1-Frg1)highReto															
		Tg(ACTA1-Frg1)lowReto															
		Tg(ACTA1-Frg1)medReto															
FRG1	Frg1																
RSPH9	Rspf9																

Tabs for Genes & Diseases information

Click to modify search

Results
You searched for:
Genes matching [Gja8 Mapk10 Rspf9 Il20ra Frg1 Jak3]

Gene Homologs x Phenotypes/Diseases [Genes (21) | Diseases (5)]

5

6

IMSR = International Mouse Strain Resource

[International Mouse Strain Resource \(IMSR\)](#)

Search for: Search Reset Show Options

You searched for: [ES Cell, embryo, live, ovaries, sperm]
Showing items 1 - 5 of 5

N	Strain Name	Synonyms	Status	Repository	Mutation Types	Aliases	Genes	Strain Types
?	CTRC1LW ^{ES}	A-Frg1 ^{ES} ES/WT1 ^{ES}	WT1 ^{ES}	ES Cell	targeted mutation	Frg1 ^{ES}	Frg1 ^{ES}	mutant strain
?	CTRC1LW ^{ES}	A-Frg1 ^{ES} ES/WT1 ^{ES}	WT1 ^{ES}	ES Cell	targeted mutation	Frg1 ^{ES}	Frg1 ^{ES}	mutant strain
?	CTRC1LW ^{ES}	A-Frg1 ^{ES} ES/WT1 ^{ES}	TGIM ^{ES}	ES Cell	gene trap	Frg1 ^{ES}	Frg1 ^{ES}	unclassified
?	SGTR ES cell line	SGTR ES cell line	MWRC ^{ES}	Other	gene trap	Frg1 ^{ES}	Frg1 ^{ES}	unclassified
?	SGTR ES cell line	SGTR ES cell line	MWRC ^{ES}	Other	gene trap	Frg1 ^{ES}	Frg1 ^{ES}	unclassified

www.findmice.org

Human Disease and Mouse Model Detail

Human Disease: Cataract 1, Multiple Types; CTRCT1
OMIM ID: 116200

Synonyms: Cataract; Cataract 1, Multiple Types, with or without Microcornea; Cataract, Duffy-Linked; Cataract, Zonular Pulverulent, 1; CZP1; C2P; CAEL

View all mouse models: View ALL (4) mouse models for this human disease.

Gene and mouse models: Mutations in human and/or mouse homologs are associated with this disease

Gene: Gja8* is associated with the disease in this species
 Mouse Homologs | Human Homologs | Mouse Models | Mouse : Human Homology Class

References: Disease References using Mouse Models (1)

In this case both the mouse gene and human ortholog are associated with the Cataract 1 disease.

Diseases tab: Disease and mouse model details

[Gene Homologs x Phenotypes/Diseases](#) [Genes \(21\)](#) [Diseases \(5\)](#)

Export: Text File

Disease	OMIM ID	Mouse Models	Associated Mouse Genes	Associated Human Genes	References using Mouse Models
Cataract 1, Multiple Types; CTRCT1	116200	4	Gja8	GJA8	3
Ciliopathy, Primary, 13	612650			RSPH9	
Fasciculohumeral Muscular Dystrophy 1; FSHD1	158900	3	Large, Tg(ACTA1-Frg1)highRot, Tg(ACTA1-Frg1)medRot, Tg(Dux4)-#Tg	DUX4, FRG1, FSHMD1A	4
Severe Combined Immunodeficiency, Autosomal Recessive, T Cell-Negative, B Cell-Positive, NK Cell-Negative	600802			JAK3	
Severe Combined Immunodeficiency, Autosomal Recessive, T Cell-Negative, B Cell-Positive, NK Cell-Negative	608971	6	Coro1a, Jak3	IL7R, PTPRC	3

<< first < prev 1 next > last >> 250

Showing results(s) 1 - 5 of 5

Human Disease and Mouse Model Detail

Human Disease: Severe Combined Immunodeficiency, Autosomal Recessive, T Cell-Negative, B Cell-Positive, NK Cell-Positive
OMIM ID: 608971

Synonyms: SCID, Autosomal Recessive, T Cell-Negative, B Cell-Positive; Severe Combined Immunodeficiency

View all mouse models: View ALL (6) mouse models for this human disease.

Gene and mouse models: Mutations in human and/or mouse homologs are associated with this disease

Gene: *Gene is associated with the disease in this species
 Mouse Homologs | Human Homologs | Mouse Models | Mouse : Human Homology Class

Coro1a* CORO1A View 4 models 1:1 Homology
 Jak3* JAK3 View 2 models 1:1 Homology
 IL7r* IL7R View 2 models 1:1 Homology
 Ptprc PTPRC View 2 models 1:1 Homology

In this case different mouse and human genes are associated with the disease.

Search by genes in VCF files

Search by genes

Ex: Bmp4, Pax*, NM_013627

Enter symbols, names or IDs. Use * for wildcard.

Upload Genes File (.txt): No file selected.

Search by genome locations

Human(GRCh38) Mouse(GRCm38)

Search by disease or phenotype terms

 Processing file and caching data matches. Please wait.

complete or continue for exact match.

GO **Reset** **BETA**

An example vcf file is available on the Dropbox site

Results

You searched for: Human locations matching [file=Pfeiffer.vcf]

Gene Homologs x Phenotype/Diseases | Genes (1741) | Diseases (263)

Legend: Terms are annotated to genes in Human Disease colors indicate more annotation.

NOTE: Search for gene or phenotype terms restricts the gene results based on the search term. Search by gene or genome location for the complete phenotype profile of gene mutations. [More...](#)

Human Gene	Mouse Gene	Phenotype/Disease
PCDH10	PCDH10	
RETN	Retrn	Rare
UBXN4	Ubxn4	Ubiquitin-like protein 4
CAZB2	Cazb2	Cazb2
NBL1	Nbl1	Nbl1
DEPPC1	Deppc1	Deppc1
EPF4C3	Epf4c3	Epf4c3
D2Z2A1	D2z2a1	D2z2a1

Search by genes in VCF files

Search by genes

Ex: Bmp4, Pax*, NM_013627

Enter symbols, names or IDs. Use * for wildcard.

Upload Genes File (.txt): No file selected.

Search by genome locations

Human(GRCh38) Mouse(GRCm38)

Search by disease or phenotype terms

VCF file successfully processed.

- 2416 Coordinates Ignored
- 3393 Rows Ignored Due to Non-Empty ID Column
- 0 Rows Ignored Due to FILTER Other Than 'PASS'
- 1634 Genes Matched On Mouse Genome
- 1741 Genes Matched On Human Genome

5830

complete or continue for exact match.

GO **Reset** **BETA**

Please verify your organism selection before hitting "GO".

OK

Search terms can be combined

Search by genome locations

Human(GRCh37) Mouse(GRCm38)

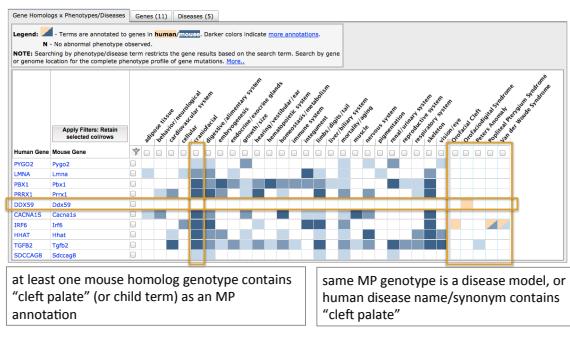
Chr1:15000000-250000000

Search by disease or phenotype terms

type "cleft palate" (matches whole)
or
type cleft palate (matches as two terms)
or select
cleft palate Mammalian phenotype
or select

Cleft Palate; CP [Cleft palate isolated, CPI] OMIM

Results for a search by both genome region and key word



Your Turn !

- Use the HMDC for a disease, gene, or phenotype you are interested in or have heard about at the Short Course
 - Try the human.vcf file on the DropBox site
 - https://www.dropbox.com/sh/seqishl631f363x/AADoBskzYjPt_IJl21dxJqV8a

Summary

- The Human-Mouse: Disease Connection supports rapid access to disease and phenotype associations
- The HMDC is in Beta release
 - contact mgi-help@jax.org with questions and suggestions