



Mouse Genome Informatics www.informatics.jax.org

The fundamental mission of the Mouse Genome Informatics resource is to facilitate the use of mouse as a model system for understanding human biology and disease.

Human Disease Models Tutorial

This tutorial will demonstrate some of the many ways to use MGI to:

- Find mouse models of human diseases
- Find mouse genotypes associated with specific phenotypes
- Find detailed phenotypic data associated with mouse alleles
- Find available mouse resources from repositories
- Obtain sets of relevant data for your own analyses

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Using the Quick Search. A Quick Search field is available on almost all MGI web pages and allows you quickly to jump to a topic of interest.

The Quick Search field appears on the top of the MGI homepage www.informatics.jax.org

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Mouse Genome Informatics

Search ▾ Download ▾ More Resources ▾ Submit Data Find Mice (IMSR) Analysis Tools Contact Us

? Keywords, Symbols, or IDs Quick Search

Explore MGI [All Search Tools](#)

Genes

 MGI Representation: Transcripts
 MGI Representation: Exons
 MGI Representation: Introns
 MGI Representation: Transcripts
 MGI Representation: Exons
 MGI Representation: Introns

Phenotypes & Disease Models

Expression

Recombinases (cre)

Function

Pathways

Strains / SNPs

Variation Type	DBP/2J	PW/NU	ROSL/PLJ	Allele Summary (all strains)
SNP	G	G	A	A/G
SNP	C	C	T	C/T

Orthology

Tumors

FAQs
 How do I...
 .. search for genes by genomic interval? [FAQ](#)
 .. find mutations for phenotypes or diseases? [FAQ](#)

News August 30, 2012
 • Gene expression data searches have been improved with new search functions, increased query performance and interactive data summaries. [Read more...](#)

The Quick Search field appears on the right side, above the navy blue navigation bar on other MGI pages

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Home Genes **Phenotypes** Expression Recombinases Function Pathways Strains / SNPs Orthology Tumors

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? Keywords, Symbols, or IDs Quick Search

Phenotypes, Alleles & Disease Models
The Phenotypes/Alleles project in MGI enables access to spontaneous, induced, and genetically-engineered mutations and their strain-specific phenotypes.

You can type *any alphanumeric string*, including **keywords** (genes, gene symbols, phenotypes, disease terms, etc.) or **accession identifiers** (from GenBank, NCBI, Ensembl, PubMed, MGI, etc.) into the Quick Search box.

In this example, the term ***papillorenal*** has been entered into the search field:

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MGI Mouse Genome Informatics

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? Quick Search

? Quick Search Results for: Search Again Reset

Examples: embryo* develop* NM_013627 MGI:97490 Fas<lpr> Pax* axial "skeletal dysplasia" Tg(ACTB)

See [details](#) for this search.

Genome Features sorted by best match, showing 1-2 of 2 [i](#)

Score	Type	Symbol	Name	Chr	Location	Str	Best Match
★★★	Chemically induced allele	Pax2^{M1Bpb}	paired box gene 2; mutation 1, Brian P Brooks	19	44830539-44912758	+	DISEASE MODEL : Papillorenal Syndrome
★★★	Targeted allele	Pax2^{tm1Pgr}	paired box gene 2; targeted mutation 1, Peter Gruss	19	44830539-44912758	+	DISEASE MODEL : Papillorenal Syndrome

Showing 1-2 of 2 [Get more data](#) for genome features 1 t

Vocabulary Terms sorted by best match, showing 1-1 of 1 [i](#)

Score	Term	Associated Data	Best Match
★★★	DISEASE Papillorenal Syndrome	5 mouse models	TERM : Papillorenal Syndrome

The Quick Search returns results in two sections:

Genome Features: matches found based on nomenclature for genes and mutations

Vocabulary Terms: matches based on diseases, phenotypes, or other keywords.

In the Genome Feature section, two mutant alleles, each annotated to the human disease term “Papillorenal Syndrome”, have been found. The allele symbols (blue) link directly to allele detail pages (see page 5) of this tutorial.

In the Vocabulary Term section, the disease term “Papilloreanal Syndrome” has been found with 5 mouse models associated. Clicking on the disease term, will lead you to the **Human Disease and Mouse Model Detail page for Papillorenal Syndrome**.

Understanding the Human Disease and Mouse Model Detail page

Human Disease and Mouse Model Detail

Human Disease	Term: Papillorenal Syndrome OMIM ID: 120330 → Online Mendelian Inheritance in Man																															
Synonyms	Coloboma of Optic Nerve with Renal Disease; Optic Coloboma, Vesicoureteral Reflux, and Renal Anomalies; Optic Nerve Coloboma with Renal Disease; Renal-Coloboma Syndrome																															
Associated Genes	Orthologous mouse and human markers where mutations in one or both species have been associated with phenotypes characteristic of this disease. <table style="width: 100%; border: none;"> <tr> <td style="width: 30%;">Mouse Gene Pax2 Gene Details</td> <td style="width: 30%;">Human Gene PAX2</td> <td style="width: 40%;">Characteristics of this human disease are associated with mutations in... ...both mouse and human orthologous genes.</td> </tr> </table>	Mouse Gene Pax2 Gene Details	Human Gene PAX2	Characteristics of this human disease are associated with mutations in... ...both mouse and human orthologous genes.																												
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Reference

The Human Disease and Mouse Model Detail page provides, for the given disease, Human Disease Name: the OMIM (Online Mendelian Inheritance in Man) disease term and a link to the OMIM record. Associated Genes: genes in *mouse and human* associated with this disease. Mouse Models: mouse genotypes published as human disease models; with links to the corresponding detail pages for the allele / phenotype data and to supporting references.

Under the Mouse Model Section of this page, the 5 unique mouse model genotypes are specified.

The following page shows a detail page for the mutant **allele Pax2^{M1Bpb}**, the first mutation (M1) in the *Pax2* gene, developed in the laboratory of Brian P Brooks (Bpb).

(note that this same detail page for *Pax2^{M1Bpb}* could have been accessed alternatively from links on the Quick Search results shown on page 3 of this tutorial) .



Allele/Phenotype Detail page for Pax2^{M1Bpb}

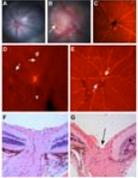
?

Pax2^{M1Bpb}

Your Input Welcome

Chemically induced Allele Detail

Nomenclature
Mutation origin
Mutation description
Find Mice (IMSR)
Phenotype summary
Phenotypes by genotype
Disease models
References

Nomenclature	<p>Symbol: Pax2^{M1Bpb}</p> <p>Name: paired box gene 2; mutation 1, Brian P Brooks</p> <p>MGI ID: MGI:4442602</p> <p>Synonyms: Pax2^{A220G}, Pax2 (p.T74A)</p> <p>Gene: Pax2 <i>Location:</i> Chr19:44830539-44912758 bp, + strand <i>Genetic Position:</i> Chr19, 38.09 cM</p>	 <p>Ocular abnormalities in Pax2^{M1Bpb}/Pax2⁺ mice</p> <p>Show the 3 image(s) involving this allele.</p>																							
Mutation origin	<p>Strain of Origin: C57BL/6</p>																								
Mutation description	<p>Allele Type: Chemically induced (ENU)</p> <p>Mutation: Single point mutation</p> <p>ENU mutagenesis induced an A to G transition at position 220 (no source sequence accession number provided) resulting in the amino acid substitution of alanine for threonine at position 74 (T74A). Reduced steady-state protein levels were confirmed by western blot analysis on embryo extracts. (J:159240)</p> <p>Inheritance: Dominant</p>																								
Find Mice (IMSR)	<p>Mouse strains and cell lines available from the International Mouse Strain Resource (IMSR)</p> <p>Carrying this Mutation: Mouse Strains: 0 strains available Cell Lines: 0 lines available</p> <p>Carrying any Pax2 Mutation: 15 strains or lines available</p>																								
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References	<p>Original: J:159240 Alur RP <i>et al.</i>, "Papillorenal syndrome-causing missense mutations in PAX2/Pax2 result in hypomorphic alleles in mouse and human." PLoS Genet 2010;6(3):e1000870</p> <p>All: 2 reference(s)</p>																								



This page contains many types of details, including characteristics of the molecular mutation, how to obtain mice carrying this mutation, specific phenotypes (expandable table, see pages 7-8 of this tutorial), images, disease models and references.

Here we look at two specific parts of this page: (1) finding mice with this mutation available from a repository and (2) examining details of the phenotypes.



(1) Finding available mice carrying *Pax2* mutations.

Find Mice (IMSR)	Mouse strains and cell lines available from the International Mouse Strain Resource (IMSR)	
	Carrying this Mutation:	Mouse Strains: 0 strains available Cell Lines: 0 lines available
	Carrying any Pax2 Mutation:	15 strains or lines available

Examine this portion of the Detail page shown on page 5 (labeled “1”). This tells us that

- 1) No mouse strains or cell lines from this particular *Pax2* mutation (*Pax2*^{M1Bpb}) are available from public repositories (but may be available from one of the publishing authors).
- 2) There are 15 strains of mice available carrying other mutations in the *Pax2* gene.

Clicking on the *15 strains or lines available* link will launch a search of the International Mouse Strain Resource (IMSR) for relevant strains from publicly available mouse repositories worldwide.

In the IMSR search results, shown below, items in the Strain Name column link to the repository's web site, the Repository column links to an e-mail or order form so you can request mice or ask about the stock and the Allele Symbol links to the MGI Allele Detail page like the one shown on page 5 of this tutorial.

International Mouse Strain Resource (IMSR)

[Search](#) [Repositories](#) [Participate](#) [Glossary](#) [Contact Us](#) [About Us](#)

Summary

Search for: [+ Show Options](#)

You searched for: << first < prev 1 next > last >> 25

Strain States: [ES Cell, embryo, live, ovaries, sperm] Showing items 1 - 15 of 15

15 strain(s) match your unfiltered search.

Export: Filter by: State ▾ Type ▾ Provider ▾ Mutation ▾

N	Strain Name	Synonyms	States	Repository	Mutation Types	Alleles
?	B6.129-Pax2^{tm1Nju}		embryo	NRCMM Order		
-	B6.129(Cg)-Pax2^{tm1Pgr/Kieg}	B6.129(Cg)-Pax2 ^{tm1Pgr} , PGR-3	embryo	EM Order	targeted mutation	Pax2^{tm1Pgr} targeted mutation 1, Peter Gruss
+	STOCK Pax2^{Opdc/H}	C3H;C-Opdc/H, Opdc(GENA380), C3H;C-Pax2 ^{Opdc/H} , STOCK Opdc/H, GENA380	embryo	EM Order	chemically induced mutation	Pax2^{Opdc} optic disc coloboma
?	CMHD ES cell line GT-276D8		ES Cell	CMMR Order	gene trap	Gt (276D8) Cmhd gene trap 276D8, Centre for Modeling Human Disease
?	CMHD ES cell line GT-287B10		ES Cell	CMMR Order	gene trap	Gt (287B10) Cmhd gene trap 287B10, Centre for Modeling Human Disease


Repository web site


Contact repository


MGI allele page

The International Mouse Strain Resource (IMSR) also can be searched directly for mouse strains of interest at <http://www.findmice.org/>.

Details of the Phenotypes

Moving to (2) examining details of the phenotypes section of the Allele Detail Page (see page 5 of this tutorial). If you are following these examples on your computer and wish to return directly to the Allele Detail page for *Pax2*^{M1Bpb}, go to: <http://www.informatics.jax.org/accession/MGI:4442602>.

The Phenotype summary section of the Allele Detail page shows a high-level view of the systems affected in mice carrying this mutation.

Phenotype summary ?	Phenotype Summary by Mammalian Phenotype terms		Key:		
	(show or hide all annotated terms)		hm homozygous	ht heterozygous	
Genotypes are listed in the next section.		cn conditional genotype	cx complex: > 1 genome feature		
		tg involves transgenes	ot other: hemizygous, indeterminate,...		
		N normal phenotype	⊗ expected model not found		
Affected Systems		Genotypes:	hm1	ht2	ht3
cardiovascular system ▶				✓	
embryogenesis ▶		✓			
mortality/aging ▶		✓			
nervous system ▶		✓	✓	✓	
pigmentation ▶			✓		
renal/urinary system ▶		✓	✓		
vision/eye ▶		✓	✓	✓	
Disease Models ▶		✓	✓		

Click on a toggle ▶ to expand a section and view more details.

A portion showing the expanded *vision/eye* section is shown below. The Disease Models section has also been toggled and shows that this allele is a model for Papillorenal Syndrome.

Phenotype summary ?	Phenotype Summary by Mammalian Phenotype terms		Key:		
	(show or hide all annotated terms)		hm homozygous	ht heterozygous	
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nervous system ▶		✓	✓	✓	
pigmentation ▶			✓		
renal/urinary system ▶		✓	✓		
vision/eye ▼		✓	✓	✓	
abnormal eye development		✓	✓		
coloboma		✓			
abnormal optic nerve morphology			✓	✓	
abnormal retina morphology			✓		
abnormal retinal blood vessel morphology			✓		
abnormal retinal blood vessel pattern			✓		
abnormal retinal pigment epithelium morphology			✓		
Disease Models ▼		✓	✓		
Papillorenal Syndrome		✓	✓		

In this grid view, a √ indicates the phenotype was detected. Each column represents a specific genotype, a combination of alleles and strain background. For details on the phenotypes shown for a specific genotype, click on an abbreviation, such as **ht2** (heterozygous genotype #2). This launches a new browser window, a portion of which is shown below.

ht2

$Pax2^{M1Bpb}/Pax2^+$

$M1Bpb$

Print

Allele combination

Strain background

vision/eye

abnormal optic nerve morphology (J:159240)

- Background Sensitivity: mice exhibit increased penetrance of optic nerve phenotype compared to mice on a mixed C3H/HeJ and C57BL/6 background
- mice exhibit excavation of the optic nerve head with prepapillary pigment changes unlike wild-type mice

Ocular abnormalities in $Pax2^{M1Bpb}/Pax2^+$ mice

abnormal eye development (J:159240)

- at E13.5, mice exhibit a delay in the formation of the tunica vasculosis lentis compared with wild-type mice
- mice exhibit variable, incomplete regression of the tunica vasculosis lentis compared with wild-type mice

abnormal retina morphology (J:159240)

- mice exhibit retinal rosettes unlike wild-type mice

abnormal retinal blood vessel morphology (J:159240)

- mice exhibit the absence of a central retinal arterial trunk unlike wild-type mice

abnormal retinal blood vessel pattern (J:159240)

abnormal retinal pigment epithelium morphology (J:159240)

- mice exhibit a mild extension of the retinal pigment epithelium beyond the borders of the optic disc unlike in wild-type mice

renal/urinary system

hydronephrosis (J:159240)

- 1 in 16 mice exhibit bilateral kidney cysts with hydronephrosis and hydroureter unlike wild-type mice

renal hypoplasia (J:159240)

- in 1 of 16 mice with contralateral double papilla
- in 1 of 16 mice with occasional focal cystic glomeruli

abnormal renal tubule morphology (J:159240)

Phenotype terms in blue

Additional descriptive text in black

Reference (number) links to publication details and abstract

Browsing for Human Diseases

We started this tutorial by searching for a human disease using the MGI Quick Search tool and drilling down for additional information based on that first search.

Another way to find human disease, syndrome and condition terms derived from the Online Mendelian Inheritance in Man (OMIM) database is to browse Human Diseases. To do this, return to the *Pax2*^{M1Bpb} Allele Detail page (<http://www.informatics.jax.org/accession/MGI:4442602>), or use any MGI page and use the “Search” pull-down menu in the navy blue navigation bar along the top.

Using the **Search** drop down, select “Vocabularies” and then “Human Disease (OMIM)”



The screenshot shows the MGI website interface. At the top, there is a search bar with the text "Keywords, Symbols, or IDs" and a "Quick Search" button. Below the search bar is a navigation bar with various tabs: Home, Genes, Phenotypes, Expression, Recombinases, Function, Pathways, Strains / SNPs, Orthology, and Tumors. A secondary navigation bar contains: Search, Download, More Resources, Submit Data, Find Mice (IMSR), Analysis Tools, and Contact Us. The main content area is titled "Pax2M1Bpb" and "Chemically induced Allele Detail". It includes a sidebar with "All Search Tools" such as Genes, Phenotypes, Expression, Recombinase (cre), Function, Pathways, Strains / SNPs, Orthology, Tumors, Sequences, References, Vocabularies, Batch Query, MouseBLAST, MGI BioMart, and Mouse Genome Browser. The "Vocabularies" section is expanded, showing options like GO Browser, Human Disease (OMIM), Mammalian Phenotype (MP) Browser, Mouse Anatomical Dictionary, and Adult Mouse Anatomy Browser. A red arrow points to the "Human Disease (OMIM)" option. The main content area displays details for the *Pax2* gene, including its location on Chromosome 19 (Chr19:44830539-44912758) and a mutation description: "paired box gene 2; mutation 1, Brian P Brooks". It also mentions "Ocular abnormalities in Pax2^{M1Bpb}/Pax2⁺ mice" and provides a link to "Show the 3 image(s) involving this allele.".

Within the Human Disease Vocabulary Browser, click any letter of the alphabet and a list of all OMIM diseases or syndromes beginning with that letter appears, arranged alphabetically (here showing the top portion of a browser page for diseases beginning with letter "S"). The number of mouse models in MGI appears in parentheses next to some disease names. The absence of this text means that MGI currently contains no mouse models for the disease.



Human Disease Vocabulary Browser

The current vocabulary contains human disease, syndrome, and condition terms from Online Mendelian Inheritance in Man ([OMIM database](#)).

Browse vocabulary terms by beginning character

[A](#) [B](#) [C](#) [D](#) [E](#) [F](#) [G](#) [H](#) [I](#) [J](#) [K](#) [L](#) [M](#) [N](#) [O](#) [P](#) [Q](#) [R](#) **[S](#)** [T](#) [U](#) [V](#) [W](#) [X](#) [Y](#) [Z](#) [0-9](#)

Human Diseases/Syndromes Beginning with "S"

To see all annotations for a disease, click the disease name.

OMIM ID Human Disease

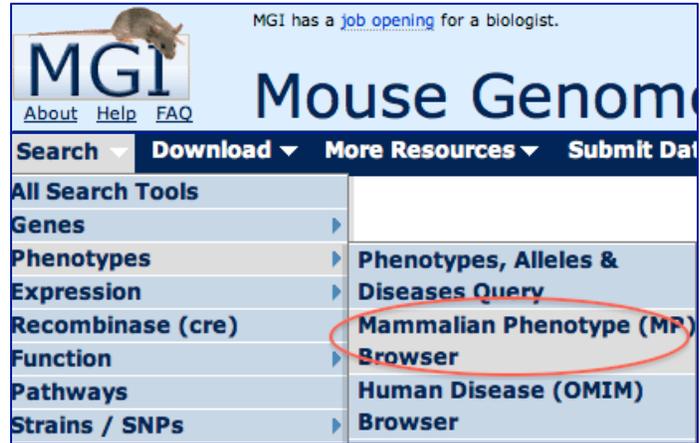
- [211390 Sabinas Brittle Hair Syndrome](#)
- [268700 Saccharopinuria](#)
- [600145 Sacral Defect with Anterior Meningocele](#)
- [101400 Saethre-Chotzen Syndrome; SCS](#) (8 mouse models)
- [610871 Sakoda Complex](#)
- [181010 Salivary Duct Calculi](#)
- [181030 Salivary Gland Adenoma, Pleomorphic](#)
- [180950 Salivary Substance, Clostridium Botulinum Type](#)
- [268800 Sandhoff Disease](#) (6 mouse models)
- [613005 Santos Syndrome](#)
- [268850 Sao Paulo MCA/MR Syndrome](#)
- [609464 Sarcoidosis, Early-Onset](#)
- [181000 Sarcoidosis, Susceptibility to, 1; SS1](#)
- [612387 Sarcoidosis, Susceptibility to, 2; SS2](#)
- [612388 Sarcoidosis, Susceptibility to, 3; SS3](#)
- [300813 Sarcoma, Synovial](#) (3 mouse models)
- [268900 Sarcosinemia](#)

The OMIM accession ID links to the OMIM database. The Human Disease term links to the MGI Human Disease and Mouse Model Detail page, like that for Papillorenal Syndrome on page 4 of this tutorial.

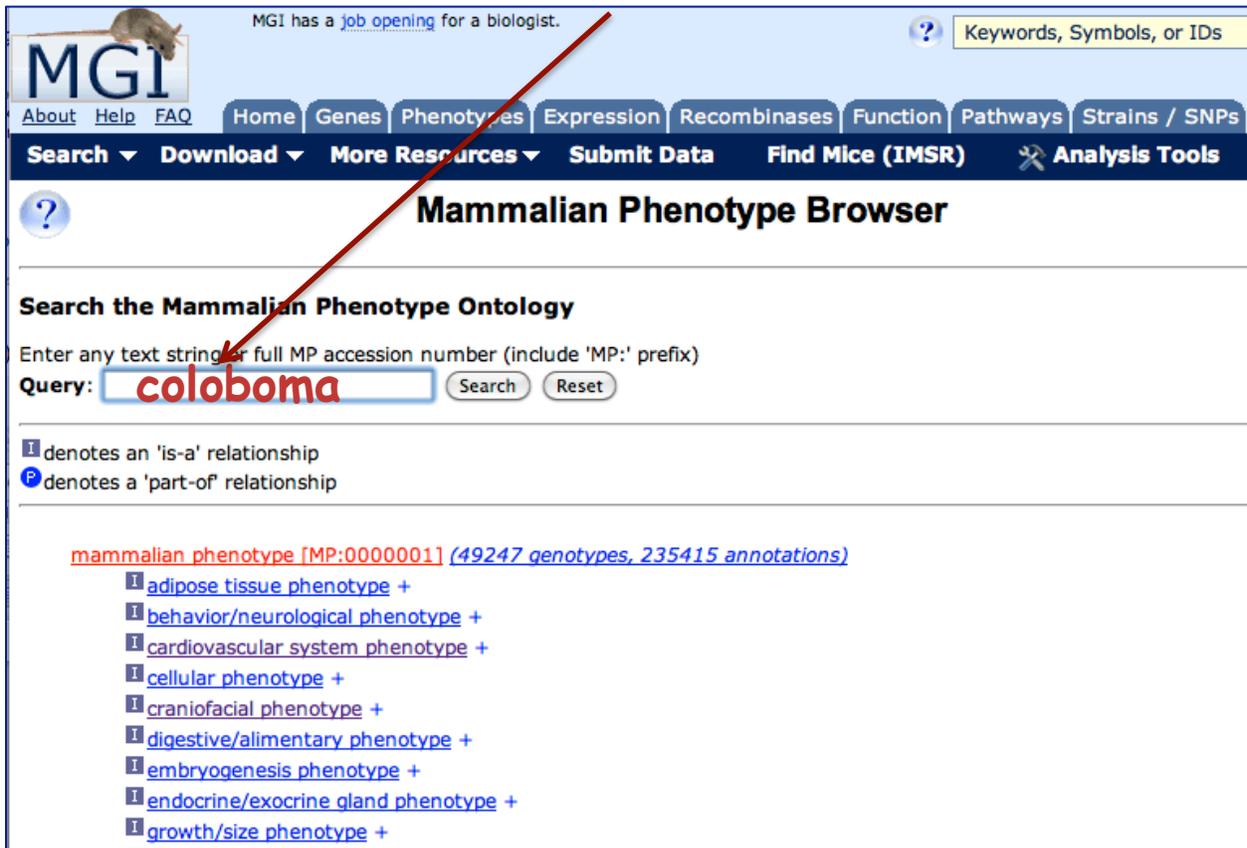
Searching by a Phenotypic Characteristic / Symptom

One of the phenotypic characteristics of Papillorenal Syndrome is *optic coloboma*. How can you find all mouse alleles associated with coloboma?

Go to the “Search” pull-down menu in the navy blue navigation bar along the top of any MGI page. Then select Mammalian Phenotype (MP) Browser from the Phenotypes section.



The Mammalian Phenotype Ontology has a hierarchical structure that permits a range of detail from high-level, broadly descriptive terms to very low-level, highly specific terms. The vocabulary can be searched or browsed. For this example, search for *coloboma*.



?

Mammalian Phenotype Browser

Query Results

7 Mammalian Phenotype term(s) matching query "coloboma":

- [choroid coloboma](#)
- [ciliary body coloboma](#)
- [coloboma](#)
- [iris coloboma](#)
- [optic disc coloboma](#)
- [optic nerve coloboma](#)
- [retina coloboma](#)

[Back to entry page](#)

This search finds 7 terms that contain the word, *coloboma*. Click on *coloboma* to find its place in the vocabulary hierarchy.

The results below for *coloboma* show the term's definition, its place in the full vocabulary hierarchy, sub-terms and a link to all the genotypes annotated to that term:

?

Mammalian Phenotype Browser

Term Detail

MP term: **coloboma**

MP id: **MP:0005262**

Alternate id: **Fyler:4311**

Definition: **anomaly in which some of the structures of the eye are absent due to incomplete fusion of the fetal intraocular fissure during gestation**

Number of paths to term: **1**

■ denotes an 'is-a' relationship
● denotes a 'part-of' relationship

mammalian phenotype

- vision/eye phenotype
 - abnormal eye morphology
 - abnormal eye development
 - abnormal corneal stroma development
 - abnormal eye muscle development
 - abnormal eyelid development +
 - abnormal lens development +
 - abnormal optic cup morphology
 - abnormal optic eminence morphology
 - abnormal optic stalk morphology
 - abnormal optic vesicle formation +
 - abnormal periocular mesenchyme morphology
 - abnormal primary vitreous morphology +
 - abnormal retinal development +
 - aniridia
 - **coloboma [MP:0005262] (62 genotypes, 68 annotations)**
 - [choroid coloboma](#)
 - [ciliary body coloboma](#)
 - [iris coloboma](#)
 - [optic disc coloboma](#)
 - [optic nerve coloboma](#)
 - [retina coloboma](#)
- cyclopia

In addition, you can access genotypes annotated to a subterm by clicking on that subterm.

Click to access the annotations

Below is a screenshot showing some of the annotations returned.

Genome-wide phenotypes

Note that the search just performed is for a *phenotype*, regardless of the genes involved; thus this search finds all mutant alleles *throughout the genome* that cause a given phenotype.

In this Annotations Page, the allele symbols (first column) link to the MGI allele detail pages, as shown previously in the *Pax2^{M1Bpb}* example on page 5 of this tutorial. The Annotated Term links to that term's place in the Mammalian Phenotype hierarchy, as in the previous figure on page 12, and the reference links to reference details.

Note that the annotations returned are for those mouse mutations where the phenotype described is *coloboma* (the searched term) or any *sub-term of coloboma*. In the example below these include phenotypes of iris coloboma, choroid coloboma, and ciliary body coloboma, which are all “types” of coloboma.

MGI has a [job opening](#) for a biologist.

Keywords, Symbols, or IDs [Quick Search](#)

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[Search](#) [Download](#) [More Resources](#) [Submit Data](#) [Find Mice \(IMSR\)](#) [Analysis Tools](#) [Contact Us](#)

Mammalian Phenotype Ontology Annotations

Query Results -- Summary

62 genotypes with 68 annotations displayed of selected term and subterms

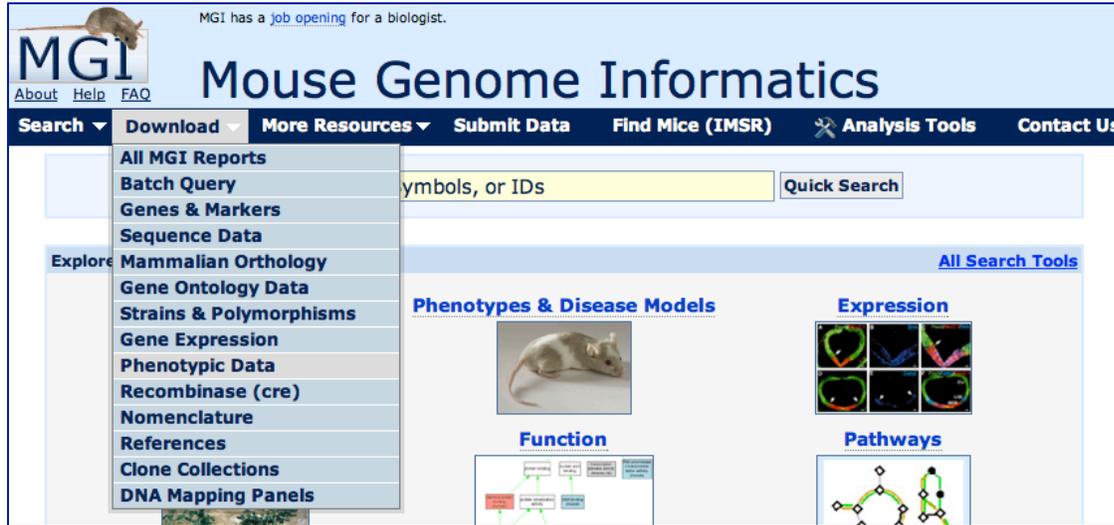
Searched Term: [coloboma](#)

Allelic Composition (Genetic Background)	Annotated Term	Reference
Cited2^{tm1Ycy}/Cited2^{tm1Ycy} (involves: C57BL/6J)	iris coloboma	J:78364
Cm/+ (involves: 101/H * C3H/HeH)	choroid coloboma	J:13451
Cm/+ Tg(Snap25)^{40Micw}/Tg(Snap25)^{40Micw} (involves: C3H/HeJ * C57BL/6J)	coloboma	J:32588
Cm/+ (C3Sn.Cg-Cm/J)	iris coloboma	J:72108
Cm/+ (Not Specified)	coloboma	J:99768
Del(2)Pax6^{11Neu}/+ (involves: 102 * C3H)	coloboma	J:151902
Del(5D5Mit388-D5Mit351)4Jcs/+ (involves: 129S4/SvJae * C57BL/6J)	iris coloboma	J:67077
Del(5D5Mit73-D5Mit351)5Jcs/+ (involves: 129S4/SvJae * C57BL/6J)	coloboma	J:67077
Ext1^{tm1Yama}/Ext1^{tm1Yama} Tg(Wnt1-cre)^{11Rth}/0 (B6.Cg-Ext1 ^{tm1Yama} Tg(Wnt1-cre) ^{11Rth})	ciliary body coloboma	J:152572
	iris coloboma	J:152572

Retrieving sets of data on Mouse Models of Human Disease

For analysis or computational aggregation, users may want sets of data rather than details of single diseases, genes, or mutations. MGI provides three avenues for retrieving data sets:

1. Use the “Download” pull-down selection of the navy blue navigation bar to access weekly generated FTP files (standard reports) that include genes/alleles/phenotypes/human disease data.



2. MGI provides a Batch Query tool where input may be genes or various IDs, and, among other data types, associated diseases and phenotypes can be retrieved and exported as Text or Excel. The Batch Query is accessed under the “Search” pull-down section from the navy blue navigation bar. The figure below shows a Bath Query Result.

Batch Summary							
Click to modify search							
Results							
You searched for:						<< first < prev 1 next > last >> 25	
Number of IDs/symbols entered: 3 Input Type: Current Symbols Only Output options: Nomenclature, OMIM 3 matching genes/markers found.							
Export: Text File Excel File							
Input	Input Type	MGI Gene/Marker ID	Nomenclature			Human Disease (OMIM) Caveat & Help	
			Symbol	Name	Feature Type	ID	Term
pax1	current symbol	MGI:97485	Pax1	paired box gene 1	protein coding gene	182940	Neural Tube Defects
pax2	current symbol	MGI:97486	Pax2	paired box gene 2	protein coding gene	120330	Papillorenal Syndrome
pax6	current symbol	MGI:97490	Pax6	paired box gene 6	protein coding gene	106210 107250 604219 148190 604229	Aniridia; AN Anterior Segment Mesenchymal Dysgenesis; ASMD Cataract, Autosomal Dominant Keratitis, Hereditary Peters Anomaly

3. The MGI BioMart tool allows selection of data filters and data attributes to produce files of customized content. Data may be exported as HTML, CSV, TSV, or Excel. Access the MGI BioMart from the “Search” pull-down section from the navy blue navigation bar. The figure below shows a BioMart Query Result.

New		Count		Results		URL	
Dataset 9778 / 84288 Entries		Export all results to		File	TSV	<input type="checkbox"/> Unique results only	Go
Genes & Genome Features		Email notification to					
Filters		View		200 rows as		HTML	<input type="checkbox"/> Unique results only
Allele Type : Targeted (knock-out)		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0000242	impaired fertilization
Attributes		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0000477	abnormal intestine morphology
Feature Symbol		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0000490	abnormal crypts of Lieberkuhn morphology
Feature Name		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0000494	abnormal cecum morphology
MGI Allele ID		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0000495	abnormal colon morphology
Allele Symbol		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0000693	spleen hyperplasia
Phenotype ID		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001127	small ovary
Phenotype Term		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001135	abnormal cervix morphology
Dataset		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001262	decreased body weight
[None Selected]		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001265	decreased body size
		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001270	distended abdomen
		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001406	abnormal gait
		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001732	postnatal growth retardation
		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001765	abnormal ion homeostasis
		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001824	abnormal thymus involution
		Cftr	cystic fibrosis transmembrane conductance regulator	MGI:1856709	Cftr^{tm1Unc}	MP:0001858	intestinal inflammation

Contact Us

We welcome questions and feedback. Contact us through the “Contact Us” link in the navy blue navigation bar at the far right of any MGI web page; or email us at mgi-help@jax.org

