



# Mouse Genome Informatics Online Resource

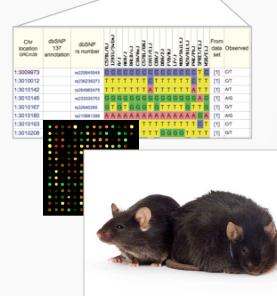
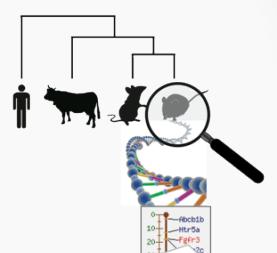
[www.informatics.jax.org](http://www.informatics.jax.org)

55<sup>th</sup> Annual Short Course on Medical and Experimental Genetics  
Joanne Berghout  
21 July 2014

[ 1 ]

## Genes, alleles and genotypes

- Within a species, all members carry the same set of **genes**
- Individual differences are due to **allelic variation**
  - “natural” background (eg. inbred line)
  - engineered variation (eg. knockout)
- Differential gene **expression** patterns allow responsiveness and differentiation.
- Phenotypes** are annotated to **genotypes**, which is a description of the total allele combination of an individual



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## Inbred laboratory mouse strains



- Laboratory mice are typically fully inbred
  - all animals are genetically identical within a strain
  - experimental consistency and reproducibility
  - differences between strains
  - comparing strains allows study of genetically influenced traits
- Individual genes can also be studied using spontaneous mutations or targeted alleles
  - MGI currently lists over 39,000 mutant alleles that have been described in mice

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## Mouse Genome Informatics



- Online resource for genes, alleles, expression and phenotypes in the laboratory mouse



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**Data:**

The screenshot shows the 'Data' section of the MGI website. It features a sidebar with icons and labels for different databases and tools, each with a small icon and a brief description. The sections include:

- Genes**: Represented by a DNA helix icon.
- Phenotypes Iomology**: Represented by a person icon.
- Strains, SNPs & Polymorphisms**: Represented by a colorful bar chart icon.
- Gene Expression Database (GXD)**: Represented by a brain and microscope icon.
- Recombinase (cre)**: Represented by a recombinase enzyme icon.
- Function**: Represented by a brain icon.

Below this, there are three more sections:

- Batch Data and Analysis Tools**: Represented by a wrench and screwdriver icon.
- Pathways**: Represented by a network diagram icon.
- Tumors**: Represented by a tissue sample icon.

At the bottom, there is a section for **Nomenclature**, represented by a flag icon.

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The screenshot shows the main homepage of the MGI website. The header includes the MGI logo, a 'CELEBRATING 25 YEARS' banner, and links for About Us, Help, FAQ, Search, Download, More Resources, Submit Data, Find Mice (IMSR), Analysis Tools, Contact Us, and Browsers.

The left sidebar contains a search bar for 'Keywords, Symbols, or IDs' and a 'Quick Search' button. Below the search bar is a list of topic-specific search tools, each with an icon:

- Genes
- Phenotypes & Mutant Alleles
- Human-Mouse: Disease Connection (BETA)
- Gene Expression Database (GXD)
- Recombinase (cre)
- Function
- Strains, SNPs & Polymorphisms
- Vertebrate Homology
- Pathways
- Tumors
- Batch Data and Analysis Tools
- Nomenclature

The right side of the page features a large banner for 'CELEBRATING 25 YEARS' with a photo of a mouse and logos for various partners. Below the banner is a 'What's new at MGI' section with a list of updates and a 'More MGI news' link. At the bottom, there are links for 'MGI Statistics' and 'More MGI news'.

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## MGI data sources



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## Outline

- Structured vocabularies, data structures in MGI
- Gene and allele navigation
- Computational and batch data access
- Translational tools

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## Structured vocabularies

- Standardized and searchable accession IDs
- Allows linking of observations from diverse experimental designs, annotating similar findings under similar headings
- Hierarchical relationships allow variable levels of precision

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## MGI's structured vocabularies:

Mammalian Phenotype Ontology Annotations		
Query Results - Summary		
Searched Term: <a href="#">abnormal brown adipose tissue physiology</a>		
MP term: 	Allieic Composition (Genetic Background)	Annotated Term Reference
MP id: 	<a href="#">Acot11<sup>tm1Deco</sup>/Acot11<sup>tm1Deco</sup></a> (involves: 12956/SvEvTac * C57BL/6)	<a href="#">abnormal brown adipose tissue physiology</a> J:182660
Definition: Number of term: 13	<a href="#">Ccdc7<sup>Tg(Prrn-Sirt1)10mai</sup>/?</a> (B6.Cg-Ccdc7 <sup>Tg(Prrn-Sirt1)10mai</sup> )	<a href="#">abnormal brown adipose tissue physiology</a> J:203814
 denotes a MP term;  denotes a MP id	<a href="#">Cebpb<sup>tm1Kish</sup>/Cebpb<sup>tm1Kish</sup></a> <a href="#">Cebpd<sup>tm1Aki</sup>/Cebpd<sup>tm1Aki</sup></a> (involves: 129P2/OlaHsd)	<a href="#">abnormal brown adipose tissue thermogenesis</a> J:45062
Definition: Number of term: 13	<a href="#">Fabp3<sup>Gt(XE705)Byg</sup>/Fabp3<sup>Gt(XE705)Byg</sup></a> (involves: 129P2/OlaHsd * C57BL/6J)	<a href="#">abnormal brown adipose tissue physiology</a> J:167535
 denotes a MP term;  denotes a MP id	<a href="#">Kdm3a<sup>tm1.1Yzha</sup>/Kdm3a<sup>tm1.1Yzha</sup></a> (B6.129P2-Kdm3a <sup>tm1.1Yzha</sup> )	<a href="#">abnormal brown adipose tissue physiology</a> J:147295
Definition: Number of term: 13	<a href="#">Lipe<sup>tm1Rze</sup>/Lipe<sup>tm1Rze</sup></a> (involves: 129P2/OlaHsd * C57BL/6J)	<a href="#">abnormal brown adipose tissue physiology</a> J:74644
 denotes a MP term;  denotes a MP id	<a href="#">Lpin1<sup>fl/fl</sup>/Lpin1<sup>fl/fl</sup></a> (BALB/cByJ-Lpin1 <sup>fl/fl</sup> )	<a href="#">abnormal brown adipose tissue physiology</a> J:63448
Definition: Number of term: 13	<a href="#">Oma1<sup>tm1Otn</sup>/Oma1<sup>tm1Otn</sup></a> (involves: 12956/SvEvTac * C57BL/6 * C57BL/6Ncr)	<a href="#">abnormal brown adipose tissue thermogenesis</a> J:184716
 denotes a MP term;  denotes a MP id	<a href="#">Plin1<sup>tm1Asg</sup>/Plin1<sup>tm1Asg</sup></a> (B6.129S6-Plin1 <sup>tm1Asg</sup> )	<a href="#">abnormal brown adipose tissue physiology</a> J:121641
Definition: Number of term: 13	<a href="#">Plin1<sup>tm1Asg</sup>/Plin1<sup>tm1Asg</sup></a> <a href="#">Tg(Fabp4-Plin1<sup>tm1Asg</sup>)</a> (involves: 12956/SvEvTac * C57BL/6)	<a href="#">abnormal brown adipose tissue physiology</a> J:121641

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## MGI's structured vocabularies:

The screenshot shows the Gene Ontology Browser interface. On the left, there are navigation links for 'Gene' and 'Molecular Function'. The main panel displays the 'Term Detail' for 'molecular\_function' (GO:0003674). The details include:

- GO term:** molecular\_function
- Synonym:** molecular function
- Synonym:** molecular function unknown
- GO id:** GO:0003674
- Definition:** Elemental activities, such as catalysis or binding, describing the actions of a gene product at the molecular level. A given gene product may exhibit one or more molecular functions.
- Comments:** Note that, in addition to forming the root of the molecular function ontology, this term is recommended for use for the annotation of gene products whose molecular function is unknown. Note that when this term is used for annotation, it indicates that no information was available about the molecular function of the gene product annotated as of the date the annotation was made; the evidence code ND, no data, is used to indicate this.
- Number of paths to term:** 1

Below the definition, there is a legend for relationship types:

- blue square: denotes an 'is-a' relationship
- blue circle: denotes a 'part-of' relationship
- green square: denotes a 'regulates' relationship
- green circle: denotes a 'positively-regulates' relationship
- red circle: denotes a 'negatively-regulates' relationship

The 'Gene\_Ontology' section lists the children of 'molecular\_function': antioxidant activity +, binding +, catalytic activity +, channel regulator activity +, chemotactant activity +, chemorepellent activity, D-alanyl carrier activity, electron carrier activity +, and enzyme regulator activity +.

The screenshot shows the Mouse Developmental Anatomy Browser interface. On the left, there is an 'Anatomy Search' sidebar where 'heart' has been entered, resulting in 17 terms sorted by best match. The results include: heart TS11-28, heart mesentery TS12-28, heart atrium TS15-28, heart ventricle TS19-28, heart septum TS17-28, heart valve TS21-28, heart blood vessel TS27-28, heart left ventricle TS19-28, heart right ventricle TS19-28, cardiac muscle tissue (heart muscle) TS12-28, cardiogenic plate (heart rudiment) TS11-12, dorsal mesocardium (dorsal heart) mesentery TS12-28, primitive heart tube TS12, early primitive heart tube TS12, early primitive heart tube cardiac muscle TS12, early primitive heart tube endocardial tube TS12, and early primitive heart tube cardiac jelly TS12.

The main panel shows the 'Anatomical Term Detail' for 'heart'. The term is highlighted in yellow. The details include:

- Term:** heart
- Present at:** Theiler Stages 11-28
- ID:** EMAPA:16105
- Synonyms:** cardium
- Parent Terms:** is-a organ, part-of cardiovascular system

On the right, there is an 'Anatomical Tree View' showing the hierarchical structure of organs and organ systems. The 'heart' node is highlighted with a red box and labeled '(32,224 expression results)'. Below the tree view, there is a summary of gene expression results:

- 13,012 genes with expression assay results
- 1,468,336 Expression assay results
- 262,508 Expression images
- 67,290 Expression assays

## Gene and allele navigation

**MGI**  
CELEBRATING 25 YEARS

**Mouse Genome Inf**

**Search ▾ Download ▾ More Resources ▾ Submit Data Find ▾**

**Apoe** Quick Search

Or use topic specific search and analysis tools:

**Genes**

**Phenotypes & Mutant Alleles**

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## Quick search ranks results by best match

Quick Search Results for: apoe Examples: embryo\* develop\* NM\_013627 MGI:97490 Fas<ipr> Pax\* axial skeletal dysplasia\* Tg(ActB-cre)2Hir

See details for this search.

<b>Genome Features</b>		sorted by best match, showing 1-10 of 179			
Score	Type	Symbol	Name	Chr	Location
★★★	protein coding gene	Apoe	apolipoprotein E	7	19696109-19699166
★★★	unclassified other genome feature	Igs7	intergenic site 7	9	Syntenic
★★★	heritable phenotypic marker	Apo	anterior polar opacity	UN	
★★★	protein coding gene	20000111101Rik	RNKEN-LDNA-2000011101 gene	13	63282143-63326096
★★★	protein coding gene	Fas	Fas (TNF receptor superfamily member 6)	19	34200659-34327770
★★★	protein coding gene	Tnfsf10	tumor necrosis factor receptor superfamily, member 25	4	152115934-152120119
★★★	protein coding gene	Tnfsf10	tumor necrosis factor (ligand) superfamily, member 10	3	27317026-27342427
★★★	Targeted allele	Apoe <sup>tm1Bres</sup>	apolipoprotein E; targeted mutation 1, Jan L Breslow	7	19696109-19699166
★★★	Targeted allele	Apoe <sup>tm1Lmh</sup>	apolipoprotein E; targeted mutation 1, Louis M Haweke	7	19696109-19699166
★★★	Targeted allele	Apoe <sup>tm1Unc</sup>	apolipoprotein E; targeted mutation 1, University of North Carolina	7	19696109-19699166

Showing 1-10 of 179 Show first 100... Get more data for genome features 1 through 179

**Vocabulary Terms** sorted by best match, showing 1-10 of 44

Score	Term	Associated Data	Best Match
★★★	FUNCTION: apolipoprotein E recycling	8 mouse models	Synonym : APOE recycling
★★★	DISEASE: Apolipoprotein E; APOE	5 genotypes, 5 annotations	TERM : Apolipoprotein E; APOE
★★★	PHENOTYPE: abnormal circulating apolipoprotein E level		Synonym : abnormal circulating ApoE level
★★★	PROTEIN DOMAIN: APO domain		TERM : APO domain
★★★	PROTEIN DOMAIN: apo-citrate lyase phosphoribosyl-dephospho-CoA transferase	9 genes, 9 annotations	TERM : apo-citrate lyase phosphoribosyl-dephospho-CoA transferase
★★★	FUNCTION: apo-beta-carotenoid-14',13'-dioxygenase activity	1 gene, 1 annotation	Synonym : APO binding
★★★	FUNCTION: death receptor binding	2 genes, 2 annotations	Synonym : Apo-1 signaling pathway
★★★	FUNCTION: Fas signaling pathway		Synonym : Apo-2L binding
★★★	FUNCTION: TRAIL binding		Synonym : apo carotenoid biosynthetic process
★★★	FUNCTION: apocarotenoid biosynthetic process		

Show all 44... 14

## Alleles, phenotypes and associated diseases

## Phenotypic allele summary

Allele Symbol Gene; Allele Name	Chr	Synonyms	Category	Abnormal Phenotypes Reported in these Systems	Human Disease Models
<i>Apoe<sup>shl</sup></i> apolipoprotein E; spontaneous hyperlipidemia	7		Spontaneous	behavior, cardiovascular, homeostasis, integument, mortality/aging	Apolipoprotein E; APOE 107741
<i>Apoe<sup>Tg(TAI)Geag</sup></i> apolipoprotein E; transgene insertion 1, George A Gaitanaris	7		Transgenic	cardiovascular, homeostasis	
<i>Apoe<sup>tm1ApoCIV</sup></i> apolipoprotein E; targeted mutation 1, Katsuhiko Yanagisawa					
<i>Apoe<sup>tm1APOCIV</sup></i> apolipoprotein E; targeted mutation 1, Shinobu C Fujita					
<i>Apoe<sup>tm1APOCIV</sup></i> apolipoprotein E; targeted mutation 1, Nobuyuki Maeda					
<i>Apoe<sup>tm1Jkw</sup></i> apolipoprotein E; targeted mutation 1, Karl H Weisgraber					
<i>Apoe<sup>tm1Jkw</sup></i> apolipoprotein E; targeted mutation 1, Jan L Breslow	7	apo E-, apoE-, EKO, epsilon-	Targeted (Null/knockout)	cardiovascular, cellular, digestive/alimentary, hematopoietic, homeostasis, immune, nervous system	
<i>Apoe<sup>tm1Jkw</sup></i> apolipoprotein E; targeted mutation 1, Karl H Weisgraber	7	<i>Apoe<sup>b</sup>, ApoelR61<sup>b</sup>, Apoe<sup>R</sup>, Arg-61<sup>new</sup></i>	Targeted (Null/knockout)	homeostasis	
<i>Apoe<sup>tm1Jkw</sup></i> apolipoprotein E; targeted mutation 1, Louis M Havekes	7	apoE-, Apoe <sup>b</sup>	Targeted (Null/knockout)	cardiovascular, homeostasis	Apolipoprotein E; APOE 107741
<i>Apoe<sup>tm1raf</sup></i> apolipoprotein E; targeted mutation 1, David L Raffai	7	Apoe <sup>b</sup>	Targeted		
<i>Apoe<sup>tm1unc</sup></i> apolipoprotein E; targeted mutation 1, University of North Carolina	7	Apoe(-), apoE-, apoE0, ApoE-KO, APOE KO, Apoe <sup>tm1unc</sup> , epsilon-, mE-, mEKO	Targeted (Null/knockout)	adipose, behavior, cardiovascular, cellular, endocrine/gocrine, growth/size, hearing/vestibular/ear, hematopoietic, homeostasis, immune, integument, liver/biliary, mortality/aging	Apolipoprotein E; APOE 107741

### Allele nomenclature:

(Gene Symbol)*alleleID* : *Apoe<sup>shl</sup>*  
 (Gene Symbol)*tm(serial number)(lab code)* : *Apoe<sup>tm1Bres</sup>*

Nomenclature   Mutation origin   Mutation description   Phenotypes   Disease models   Find Mice (IMSR)   Notes   References																																																																																																										
<b>Nomenclature</b>	<b>Symbol:</b> <i>Apoe<sup>shl</sup></i> <b>Name:</b> apolipoprotein E; spontaneous hyperlipidemia <b>MGI ID:</b> MGI:3574899 <b>Gene:</b> <i>Apoe</i> <b>Location:</b> Chr7:19696109-19699166 bp, - strand <b>Genetic Position:</b> Chr7, 9.94 cM					 Show the 2 image(s) involving this allele.																																																																																																				
<b>Mutation origin</b>	Strain of Origin: KOR																																																																																																									
<b>Mutation description</b>	<b>Allele Type:</b> Spontaneous <b>Mutation:</b> Undefined <small>Mutation details: This mutation was shown to be an allele of Apoe by complementation testing against Apoe<sup>tm1unc</sup> and by observation of altered restriction fragment sizes upon Southern blot analysis using a partial Apoe cDNA probe. (J:54051)</small>																																																																																																									
<b>Phenotypes</b>	<b>Inheritance:</b> Recessive <b>Key:</b> hm: homozygous    ht: heterozygous    ct: complex: > 1 genome feature    tg: involves transgenes    o: other: hemizygous, indeterminate,...    n: normal phenotype																																																																																																									
	<b>Genotypes:</b> <table border="1"> <thead> <tr> <th>Genotype</th> <th>Allelic Composition</th> <th>Genetic Background</th> <th>Cell Line(s)</th> </tr> </thead> <tbody> <tr> <td>hm1</td> <td>Apoe<sup>shl</sup>/Apoe<sup>shl</sup></td> <td>B6.KOR-Apoe<sup>shl</sup></td> <td></td> </tr> <tr> <td>hm2</td> <td>Apoe<sup>shl</sup>/Apoe<sup>shl</sup></td> <td>C.KOR-Apoe<sup>shl</sup></td> <td></td> </tr> <tr> <td>hm3</td> <td>Apoe<sup>shl</sup>/Apoe<sup>shl</sup></td> <td>C3.KOR-Apoe<sup>shl</sup></td> <td></td> </tr> <tr> <td>Disease Model</td> <td>Apoe<sup>shl</sup>/Apoe<sup>shl</sup></td> <td>involves: BALB/c * KOR</td> <td></td> </tr> <tr> <td>hm5</td> <td>Apoe<sup>shl</sup>/Apoe<sup>shl</sup></td> <td>involves: C3H/He * KOR</td> <td></td> </tr> <tr> <td>Disease Model</td> <td>Apoe<sup>shl</sup>/Apoe<sup>shl</sup></td> <td></td> <td></td> </tr> </tbody> </table> <b>Phenotypes:</b> <b>Affected Systems</b> <table border="1"> <thead> <tr> <th>Sex</th> <th>hm1</th> <th>ht</th> <th>hm2</th> <th>hm3</th> <th>tg</th> <th>o</th> <th>hm5</th> <th>ct</th> <th>?</th> <th>♀</th> <th>♂</th> </tr> </thead> <tbody> <tr> <td>behavior/neurological</td> <td>▶</td> <td></td> <td></td> <td></td> <td></td> <td></td> <td></td> <td></td> <td></td> <td>✓</td> <td></td> </tr> <tr> <td>cardiovascular system</td> <td>▶</td> <td>✓</td> <td>✓</td> <td>✓</td> <td>✓</td> <td>✓</td> <td>✓</td> <td>✓</td> <td></td> <td></td> <td></td> </tr> <tr> <td>homeostasis/metabolism</td> <td>▶</td> <td>✓</td> <td>✓</td> <td>✓</td> <td>✓</td> <td>✓</td> <td>✓</td> <td>✓</td> <td></td> <td></td> <td></td> </tr> <tr> <td>integument</td> <td>▶</td> <td></td> <td></td> <td></td> <td>✓</td> <td>✓</td> <td>✓</td> <td></td> <td></td> <td></td> <td></td> </tr> <tr> <td>mortality/aging</td> <td>▶</td> <td></td> <td></td> <td></td> <td>✓</td> <td>✓</td> <td>✓</td> <td></td> <td></td> <td></td> <td></td> </tr> </tbody> </table>					Genotype	Allelic Composition	Genetic Background	Cell Line(s)	hm1	Apoe <sup>shl</sup> /Apoe <sup>shl</sup>	B6.KOR-Apoe <sup>shl</sup>		hm2	Apoe <sup>shl</sup> /Apoe <sup>shl</sup>	C.KOR-Apoe <sup>shl</sup>		hm3	Apoe <sup>shl</sup> /Apoe <sup>shl</sup>	C3.KOR-Apoe <sup>shl</sup>		Disease Model	Apoe <sup>shl</sup> /Apoe <sup>shl</sup>	involves: BALB/c * KOR		hm5	Apoe <sup>shl</sup> /Apoe <sup>shl</sup>	involves: C3H/He * KOR		Disease Model	Apoe <sup>shl</sup> /Apoe <sup>shl</sup>			Sex	hm1	ht	hm2	hm3	tg	o	hm5	ct	?	♀	♂	behavior/neurological	▶									✓		cardiovascular system	▶	✓	✓	✓	✓	✓	✓	✓				homeostasis/metabolism	▶	✓	✓	✓	✓	✓	✓	✓				integument	▶				✓	✓	✓					mortality/aging	▶				✓	✓	✓					
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	<a href="#">View phenotypes for all genotypes (concatenated display).</a>																																																																																																									
<b>Disease models</b>	<b>Key:</b> ✓ disease model    Ⓢ expected model not found <b>Models:</b> Human Diseases Apolipoprotein E; APOE OMIM: 107741																																																																																																									
<b>Find Mice (IMSR)</b>	Mouse strains and cell lines available from the International Mouse Strain Resource (IMSR) Carrying this Mutation: Mouse Strains: 1 strain available    Cell Lines: 0 lines available Carrying any Apoe Mutation: 57 strains or lines available																																																																																																									
<b>Notes</b>	Northern blot analysis of liver RNA and immunoblot analysis of plasma from homozygous mice demonstrated lack of expression of Apoe RNA and protein. The phenotype is generally similar to that of targeted Apoe mutations on the same diet. Mice of the original mutant strain (SHL) exhibit more severe xanthoma and hypercholesterolemia, and at an earlier age, than do homozygous mice congenic for the mutation on three standard laboratory inbred-strain backgrounds. In contrast, atherosclerotic lesions of the aortae of congenic mutants are more extensive than those of SHL mice. SHL mice seldom survive past 12 months of age.																																																																																																									
<b>References</b>	<b>Original:</b> J:54051 Matsushima Y, et al., Spontaneously hyperlipidemic (SHL) mice: Japanese wild mice with apolipoprotein E deficiency. <i>Mamm Genome.</i> 1999 Apr;10(4):352-7 <b>All:</b> 5 reference(s)																																																																																																									

## Phenotype tables annotate observations to genotypes

**Key:**

hm	homozygous
cn	conditional

**Genotypes:**

hm1	Apoe <sup>shl</sup>	Apoe <sup>shl</sup>
hm2	hm2	C.KOR-Apoe <sup>shl</sup>
hm3		
hm4	Disease	
hm5	Disease	

**Key:**

♀	phenotype observed in females	WTSI	Wellcome Trust Sanger Institute
♂	phenotype observed in males	EuPh	Europhenome
N	normal phenotype		

**Print**

**homeostasis/metabolism**

increased circulating cholesterol level ( J:75488 )

- Background Sensitivity: cholesterol levels in various genetic backgrounds are as follows (highest to lowest levels): KOR-Apoe<sup>shl</sup>, C3.KOR-Apoe<sup>shl</sup>, C.KOR-Apoe<sup>shl</sup>, and B6.KOR-Apoe<sup>shl</sup>
- Background Sensitivity: cholesterol levels increase with age on a BALB/c background

xanthoma ( J:75488 )

- Background Sensitivity: lesions are hardly apparent on a BALB/c background

**cardiovascular system**

increased susceptibility to atherosclerosis ( J:75488 )

- Background Sensitivity: increased susceptibility varies based on background strain as follows (most prominent to least prominent): B6.KOR-Apoe<sup>shl</sup>, C.KOR-Apoe<sup>shl</sup>, C3.KOR-Apoe<sup>shl</sup>, KOR-Apoe<sup>shl</sup>

	increased susceptibility to atherosclerosis	homeostasis/metabolism	integument	mortality/aging
increased susceptibility to atherosclerosis	✓	✓	✓	✓
homeostasis/metabolism	►	✓	✓	✓
integument	►		✓	✓
mortality/aging	►		✓	✓

[View phenotypes for all genotypes \(concatenated display\).](#)

## Genes and Alleles Section Summary

- MGI provides data and tools for the research community in a relational database
- Genes, alleles and phenotypes are described using searchable, structured terms as well as more detailed free text
  - each piece of information is cited with a J:# referring back to the source of the information

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## Outline

- Structured vocabularies
- Gene and Allele navigation
- Computational access
- Translational tools

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## Computational access

**Batch Data and Analysis Tools**

[Batch Queries](#) | [Data Files for Download](#) | [Analysis Tools](#)

**Batch Queries**

- [Batch Query](#)  
Input a list of IDs or gene symbols and retrieve other database IDs and gene attributes (e.g. phenotypes, GO, expression data).
- [MouseMine](#)  
Powered by InterMine, MouseMine offers flexible querying, numerous predefined query templates, iterative refinement of results, and linking to other model organism Mines.  
MouseMine:
  - offers many useful "canned" queries, plus point-and-click query editing
  - supports uploading and manipulating lists of objects
  - allows any query result to be downloaded (text, xml, json) or forwarded to Galaxy
- [MGI BioMart](#)  
Access MGI's mouse genetic, genomic and expression data using BioMart.

**Data Files for Download**

**Public Reports via FTP**

- Over 75 data files are generated weekly and available for download.
- See [MGI Data and Statistical Reports](#) for the list of reports and their field descriptions.

**Tab Delimited Output from our Web Interface**

- User-defined results for references, gene and SNP data are available in tab delimited format.
- You can modify the output format on the query form from *Web* to *Tab-delimited*.
- Query Forms with the tab-delimited option available are:
  - [MGI Batch Query](#)  
Download gene/marker data for a batch of IDs or symbols
  - [Genes and Markers Query Form](#)  
Search by symbol, location, gene ontology classification, or phenotype.
  - [Mouse SNP Query Form](#)

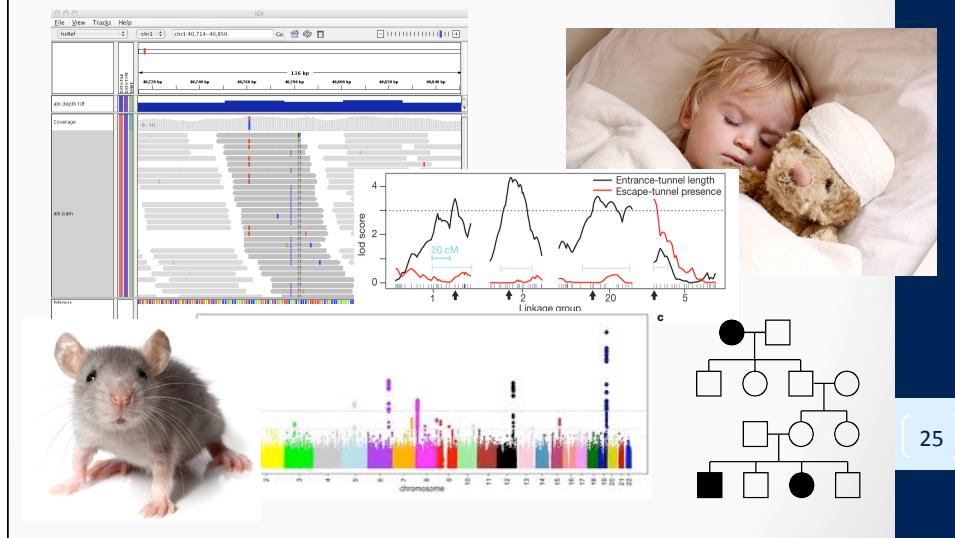
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## Computational and Batch Data Section Summary

- MGI generates daily and weekly tabular reports of data
- MouseMine allows straightforward, flexible batch-scale querying of MGI data
- MouseMine contains multiple useful list analysis tools
  - enrichment analysis
  - intersections with other MGI data

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# Clinical genetic research and translation



**Human Mouse: Disease Connection**

Relating human diseases and mouse models

**Search by genes**

Ex: *Bmp4*, *Pax\**, NM\_013627

**Search by genome locations**

Ex: Chr1:3000000-10000000

**Search by disease or phenotype terms**

Ex: *diabetes*, 105830

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

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

**Upload a VCF File:** Browse... No file selected.  Human(GRCm37)  Mouse(GRCm38)

**GO** **Reset** **BETA**

**Human-Mouse: Disease Connection**

Take a tour of the Human-Mouse: Disease Connection

**Spotlight on mouse models of human disease**

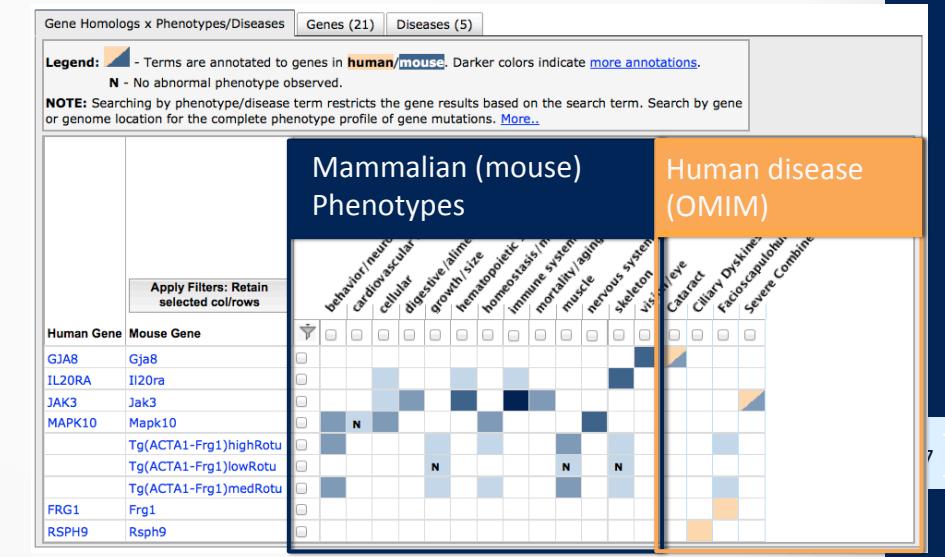
**Human T-Cell Immunodeficiency, Congenital Alopecia, and Nail Dystrophy (OMIM: 601705)**

Humans and mice homozygous for recessive mutations in the FOXN1 (forkhead box N1) gene display common phenotypes:

- congenital alopecia
- absent thymus
- severe T-cell immunodeficiency
- nail dystrophy
- limited lifespan

[Read more...]

## Visual display of associated phenotype and disease results



## Translational tools Section Summary

- The **Human-Mouse: Disease Connection** allows rapid accession and association of gene-phenotype or gene-disease information
  - Allows clinical researchers with human data to perform functional or phenotypic annotation to a gene list

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## Questions?

- See: [MGI Practical Worksheet](#) link on SharePoint in the Introduction to Bioinformatics folder
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