

Accessing the Mouse Genome: Genome Variation

IMGC Bioinformatics Tutorial
Charleston, South Carolina
November 12, 2006

Carol Bult, The Jackson Laboratory

Deanna Church, NCBI



Tutorial Aims

The primary aims of this tutorial are to demonstrate how to use publicly available bioinformatics resources to query for and access mouse SNP data according to:

- Strains of interest
- Genome/chromosome location
- Gene of interest
- SNP function class
- Associations with phenotype measurement data

Resources covered in this tutorial

The two primary resources used for this tutorial are the Mouse Genome Informatics (MGI) database and the Mouse Phenome Database (MPD).

- **MGI** draws data and information from the primary scientific literature and from other database resources including NCBI, Ensembl, and VEGA. These other database resources, in turn, take information from MGI and incorporate this information into their resources. For example, many sites display official mouse gene and strain nomenclature, genetic mapping data, gene functional annotations, phenotype annotations, and gene expression data that are obtained from MGI.
- **MPD** is a database of phenotype measurement data and genotype data for the laboratory mouse. The database supports a specific community initiative, the Mouse Phenome Project, to systematically collect phenotype measurements for commonly used inbred mice using methods that allow for robust statistical analysis and comparisons among strains of mice.

Sources of mouse SNP data

The SNP data in the Mouse Genome Informatics (MGI) database is downloaded from NCBI's dbSNP database. The SNP data in MGI are refreshed when there is a new public release of dbSNP.

The Mouse Phenome Database (MPD) downloads SNP data from dbSNP. MPD also loads SNP data from data providers prior to the release of these data via dbSNP.


MGI_3.44 - Mouse Genome Informatics - Microsoft Internet Explorer

File Edit View Favorites Tools Help

Back Forward Stop Home Search Favorites

Address http://www.informatics.jax.org/

The MGI Home Page
<http://www.informatics.jax.org>



Mouse Genome Informatics
 Mouse Genome Informatics (MGI) provides integrated access to data on the genetics, genomics, and biology of the laboratory mouse.

Search for
 Enter term(s) or part of a term, use * as a wildcard.

in these sections

- All sections
- Gene symbols/names
- Accession IDs
- Phenotype/Human Disease
- Gene Expression

Search
Reset

Advanced search for...

Search Menus
 Links to advanced search forms, database reports...

- [All Search Tools](#)
- [Genes and Markers](#)
- [Phenotypes and Alleles](#)
- [Strains and Polymorphisms new!](#)
- [Gene Expression](#)
- [Sequences](#)
- [Mammalian Orthology and Comparative Maps](#)
- [Mouse Maps and Mapping Data](#)
- [Mouse Tumor Biology](#)
- [Probes and Clones](#)

[What's New](#)
 MGI news and announcements

[Additional MGI Tools and Links](#)
 Nomenclature, online books, E-mail lists, ftp server, prototypes...

[Data Submission](#)
 Instructions and tools for submitting your data

start Microsoft PowerPoint PGA_Tutorial_2006 - ... MGI_3.44 - Mouse Ge... Internet 1:55 PM

me to the Mouse Phenome Database - Microsoft Internet Explorer

Home Hints FAQ Contents Tools SNPs Downloads Preferences About Search

The MPD Home Page
<http://www.jax.org/phenome>

Mouse Phenome Database Your flagged measurements

Welcome to the Mouse Phenome Database

...a collection of phenotypic and genotypic data for the laboratory mouse

What's new

About MPD

Downloads

Recommendations

MPD priority strains

Data submission guidelines

Assays & completion status

Funding

Steering committee

Search MPD: [Help](#)

Tools Hints FAQ

Measurements

- anatomy
- appearance
- behavior
- biological factors
- blood
- cancer
- diet effects
- drug effects, toxicity
- genotype
- heart, lung
- intake, metabolism
- musculoskeletal
- neurosensory
- reproduction
- other / all

Mouse SNPs Updated Build 36

Projects Protocols Participants

Mouse strains

- MPD priority strains
- Other strains: recombinant inbred, chromosome substitution strains, mutant strains, others

When mentioning the MPD please use this URL: <http://www.jax.org/phenome> Email: phenome@jax.org

Spidering of the MPD web site is restricted. Use downloads instead.

Exercise 1: Finding all SNPs in a chromosomal region for one or more strains in MGI

1. Go to the MGI home page
 - a. <http://www.informatics.jax.org>
2. Click on the **Strains and Polymorphisms** search tool menu link
3. Select the **SNPs query form**
4. Select any number of strains from the *Available Strains* panel and move them into the *Selected Strains* panel using the arrows
5. Select a *Chromosome* or chromosome region
6. Click on the **Search** button in the top left corner of the query form
 - a. A new window will open up that will display your search results. The query form will stay open so that you can modify your search parameters without using your web browser's "Back" button.

1. Strain Selection

2. Chromosome Selection

Exercise 1; Steps 4, 5

Mouse SNPs
Query Results -- Summary

You searched for...
Selected Strain(s): equals 129S1/SvImJ or 129X1/Sv or C3H/HeJ or C57BL/6J
Chromosome: equals 14
Display Limit: equals 1000
Allele Display Mode: equals nucleotide values

1000 of 228591 matching SNPs displayed

Legend
Results are sorted by chromosome/coordinate value. Chromosome transitions are marked by blank rows.

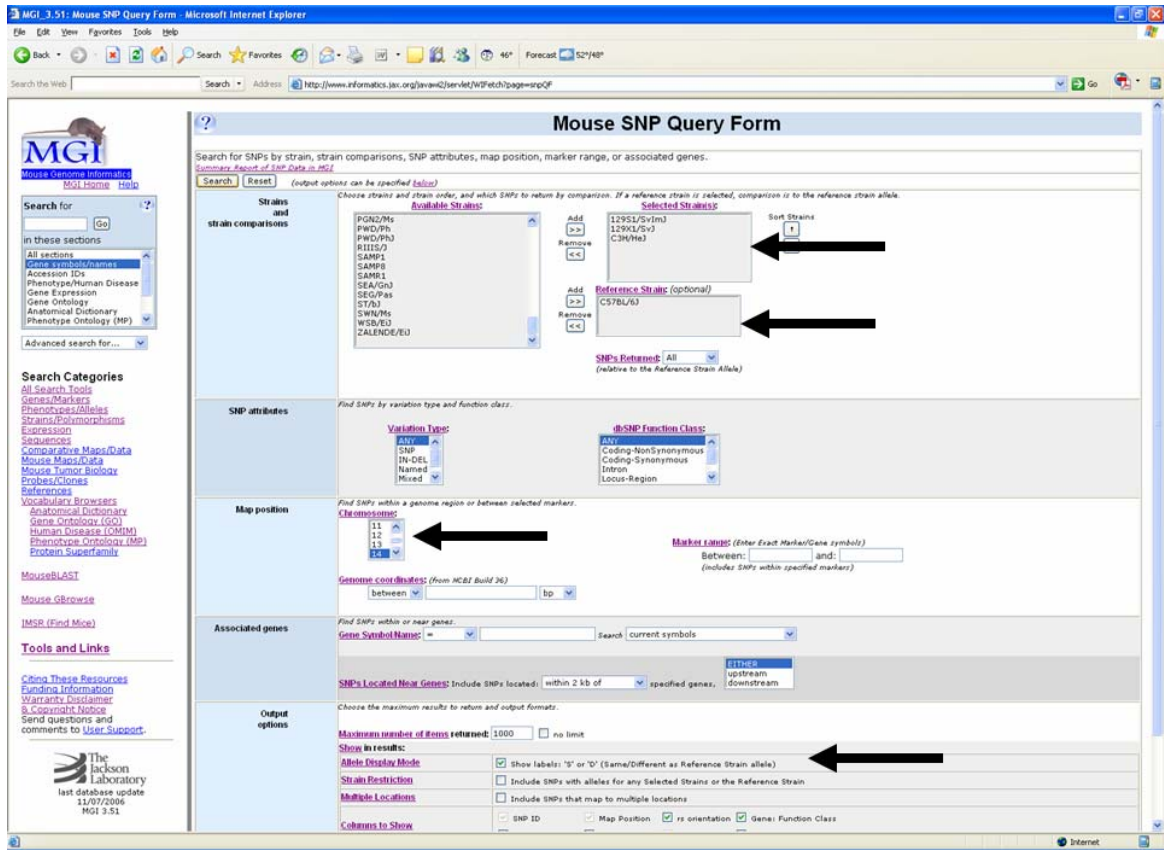
SNP ID	Map Position (NCBI Build 36)	Orientation	Gene : dbSNP Function Class	Assays (ss)	Variation Type	129S1/SvImJ	129X1/Sv	C3H/HeJ	C57BL/6J	Allele Summary (all strains)
rs30674430 MED dbSNP MGI SNP Detail	Chr14:3114869	f		1	SNP				A	A/G
rs36549200 MED dbSNP MGI SNP Detail	Chr14:3668891	f		1	SNP	C	T			C/T
rs37209719 MED dbSNP MGI SNP Detail	Chr14:3669024	f		1	SNP	C				C/G
rs37209317 MED dbSNP MGI SNP Detail	Chr14:3669317	f		1	SNP	A	A			A/G
rs36739445 MED dbSNP MGI SNP Detail	Chr14:3669380	f		1	SNP	A	T			A/T
rs37253052 MED dbSNP MGI SNP Detail	Chr14:3669937	f		1	SNP	T	C			C/T
rs37527296 MED dbSNP MGI SNP Detail	Chr14:3670035	f		1	SNP	A	A			A/G
rs38141919 MED dbSNP MGI SNP Detail	Chr14:3670160	f		1	SNP	C	C			C/T
rs36456330 MED dbSNP MGI SNP Detail	Chr14:3671357	f		1	SNP	C	C			C/G
rs36384420 MED dbSNP MGI SNP Detail	Chr14:3671948	f		1	SNP	A	A			A/G
rs36727553 MED dbSNP MGI SNP Detail	Chr14:3672185	f		1	SNP	A	A			A/T
rs37518632 MED dbSNP MGI SNP Detail	Chr14:3677484	f		1	SNP	C	G			C/G
rs36496266 MED dbSNP MGI SNP Detail	Chr14:3678043	f		1	SNP	A	A			A/G
rs37129290 MED dbSNP MGI SNP Detail	Chr14:3678381	f		1	SNP	A	A			A/T
rs36787325 MED dbSNP MGI SNP Detail	Chr14:3678388	f		1	SNP	C	C			C/T
rs37515454 MED dbSNP MGI SNP Detail	Chr14:3682877	f		1	SNP	T	G			G/T
rs37097874 MED dbSNP MGI SNP Detail	Chr14:3691578	f		1	SNP	C	C			A/G
rs39191645 MED dbSNP MGI SNP Detail	Chr14:3694339	f		1	SNP	A	A			A/G
rs36851917 MED dbSNP MGI SNP Detail	Chr14:3698793	f		1	SNP	T	A			A/T
rs3731283 MED dbSNP MGI SNP Detail	Chr14:3699117	f		1	SNP	A	G			A/G

Query results show all available SNPs for the selected strains, not just SNPs relative to a Reference Strain.

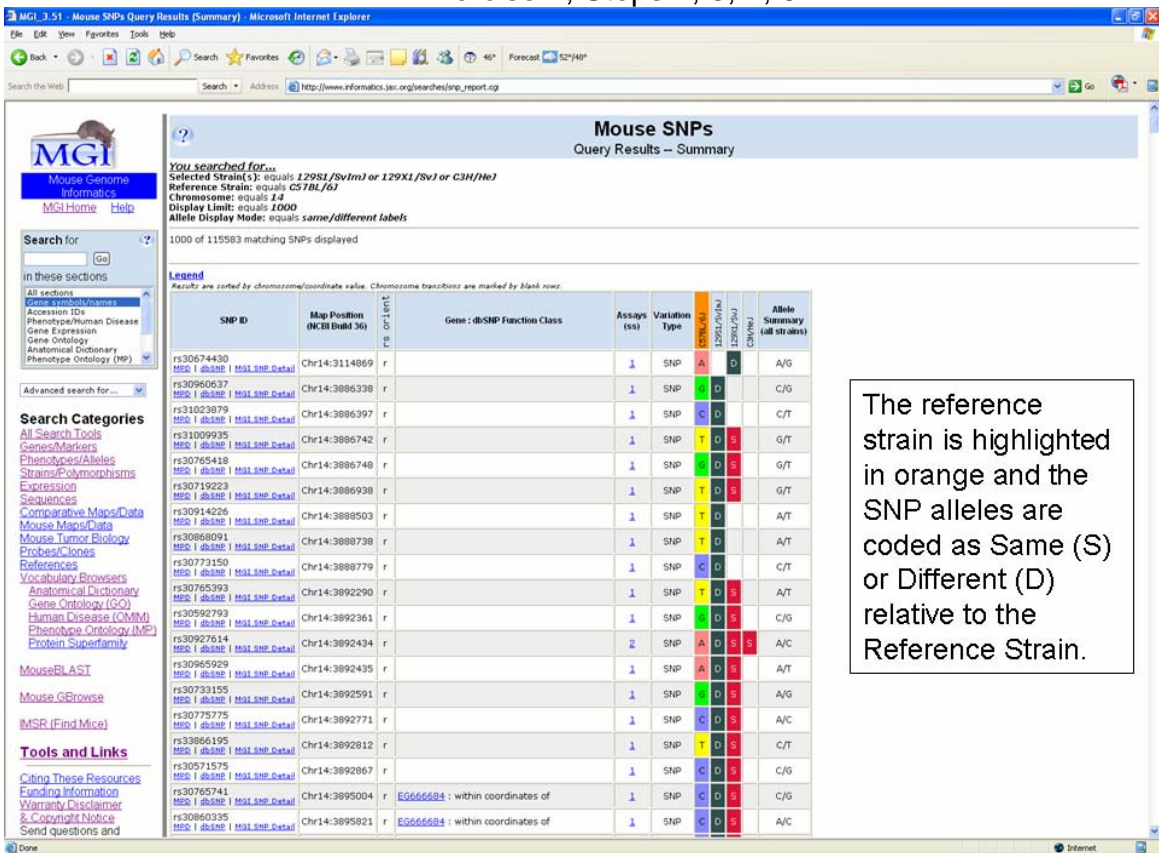
Exercise 1; Step 6 results

Exercise 2: Finding all SNPs in a chromosomal region for one or more strains relative to a Reference Strain

- Return to the **SNP query form** from Exercise 1. Your query parameters from Exercise 1 should still be active in the query form.
- Highlight the **C57BL/6J** strain name in the *Selected Strains* panel. Then move it back it to the *Available Strains* list using the arrow keys.
 - When you move the strain name back it will go to the bottom of the Available Strains list
- Select **C57BL/6J** from the *Available Strains* list and move it to the *Reference Strain* panel using the arrow keys.
- Be sure you have the same chromosome or chromosome region selected as you did in Exercise 1.
- At the bottom of the **SNP query form** in the *Output Options* section, select the option to display the SNP alleles as "Same" or "Different" relative to the selected Reference Strain
- Click on the **Search** button in the top left corner of the query form
 - If you have not closed your original query results window from Exercise 1, it will be refreshed with the new query results.



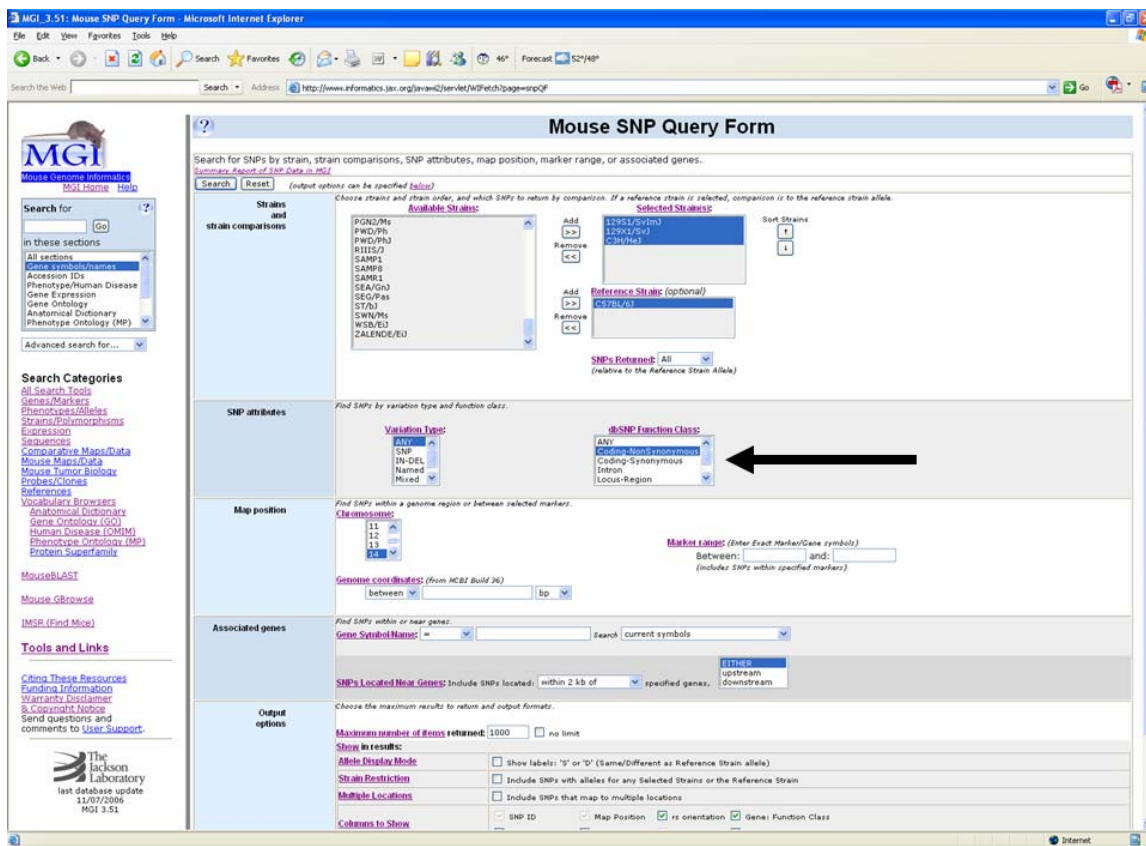
Exercise 2; Steps 2, 3, 4, 5



Exercise 2; Step 6 results

Exercise 3: Querying for specific SNP function classes

1. Return to the **SNP query form** from Exercise 2. Keep the parameters from Exercise 2.
2. In the **SNP attributes** section of the query form, go to the **dbSNP Function Class** and select “Coding Non-Synonymous”.
 - a. It is possible to select more than one Function Class at a time
3. At the bottom of the **SNP query form** in the **Output Options** section, de-select the option to display the SNP alleles as “Same” or “Different” relative to the selected Reference Strain
4. Click on the **Search** button in the top left corner of the query form
5. In the query results page, explore some of the hyper text links on the page
 - a. The gene symbol links will take you to gene detail pages in MGI
 - b. The MGI SNP detail page will take you to SNP details
 - c. The dbSNP link should take you to the refSNP record in dbSNP but there has been a problem with this link due to a bug at dbSNP.
 - d. The MPD link will take you to SNP details at the Mouse Phenome Database (MPD)



Exercise 3; Step 2

MGI 3.51: Mouse SNPs Query Results (Summary) - Microsoft Internet Explorer

Search for: Go

Address: http://www.informatics.jax.org/searches/snps_report.cgi

Mouse SNPs Query Results -- Summary

You searched for...
 Selected Strain(s): equals 12981/SvEv or 129X1/SvEv or C3H/HeJ
 Reference Strain: equals C57BL/6J
 Function Class: equals Coding-NonSynonymous
 Chromosome: equals 14
 Display Limit: equals 2000
 Allele Display Mode: equals nucleotide values

338 matching SNPs displayed

Legend
 Results are sorted by chromosome/coordinate value. Chromosome transitions are marked by blank rows.

SNP ID	Map Position (NCBI Build 36)	rs or dbSNP	Gene: dbSNP Function Class	Assays (ss)	Variation Type	12981/SvEv	129X1/SvEv	C3H/HeJ	C57BL/6J	Allele Summary (all strains)
rs131062101	Chr14:6955557	f	Abca2 : Coding-NonSynonymous	1	SNP	T	C	C	C	C/T
rs3097979	Chr14:7043734	c	Abca2 : Coding-NonSynonymous	1	SNP	T	A	A	A	A/T
rs30320290	Chr14:11077805	f	Abca2 : Coding-NonSynonymous	1	SNP	C	A	A	A	C/G
rs13482063	Chr14:12571146	f	Abca2 : Coding-NonSynonymous	2	SNP	A	G	G	G	A/G
rs30431141	Chr14:12707692	f	Gm291 : Coding-NonSynonymous	1	SNP	G	A	A	A	A/G
rs30667631	Chr14:12707694	f	Gm291 : Coding-NonSynonymous	1	SNP	T	C	C	C	C/T
rs1279232	Chr14:12806896	f	Abca7 : Coding-NonSynonymous	1	SNP	A	A	A	A	A/G
rs13482064	Chr14:12893837	f	Abca7 : Coding-NonSynonymous	2	SNP	A	C	C	C	A/C
rs4230191	Chr14:15035538	f	Qsox1 : Coding-NonSynonymous	1	SNP	A	A	A	A	A/T
rs3685768	Chr14:17900858	f	Egfr32498 : Coding-NonSynonymous	1	SNP	A	A	A	A	A/G
rs31328987	Chr14:17901141	f	Egfr32498 : Coding-NonSynonymous	1	SNP	C	T	T	T	C/T
rs30667034	Chr14:18580468	f	Nid2 : Coding-NonSynonymous	2	SNP	A	A	A	A	A/G
rs13136598	Chr14:19237141	f	Tbc18 : Coding-NonSynonymous	1	SNP	A	A	A	A	A/G
rs30306452	Chr14:19253492	f	Anxa7 : Coding-NonSynonymous	2	SNP	A	C	A	A	A/C
rs30691085	Chr14:19475118	f	Sec24c : Coding-NonSynonymous	2	SNP	A	A	A	A	A/G
rs30370500	Chr14:19481329	f	Sec24c : Coding-NonSynonymous	1	SNP	A	C	C	C	C/G
rs30306736	Chr14:20776019	f	Zfp593 : Coding-NonSynonymous	1	SNP	C	T	T	T	C/T
rs30286470	Chr14:25502427	f	E430028219a : Coding-NonSynonymous	1	SNP	C	T	T	T	C/T
rs30256712	Chr14:25502427	f	E430028219a : Coding-NonSynonymous	1	SNP	A	T	T	T	A/T

Exercise 3; Step 2

MGI 3.51: Gene Detail - Microsoft Internet Explorer

Search for: Go

Address: http://www.informatics.jax.org/branch2/gen-let/W?PetCh=page=markerDetail&key=64356

Gene Detail

Symbol: **Acox2**
 Name: **acyl-Coenzyme A oxidase 2, branched chain**

Genetic Map: Chromosome 14, cytoband A1, Mapping data(1)

Sequence Map: Chr14:7019267-7052692 bp, - strand (From NCBI annotation of NCBI Build 36)

Mammalian homology: human; rat (Mammalian Orthology), Protein Superfamily: [acyl-CoA oxidase](#)

Sequences:

Representative Sequences	Length	Strain/Species	Flank
<input type="checkbox"/> genomic 93732		C57BL/6J	a 0 kb
<input type="checkbox"/> transcript NM_053115	2499	-	-
<input type="checkbox"/> polypeptide Q9QXD1	681	Not Applicable	-

Polymorphisms: SNPs within 2kb(25)

Gene Ontology:

- Process: [electron transport, fatty acid beta-oxidation...](#)
- Component: [peroxisome](#)
- Function: [acyl-CoA dehydrogenase activity, acyl-CoA oxidase activity...](#)

Expression:

Therier Stage	15	19	22	26	28	Tissues(9)	Results(15)	Assays(1)
Assay Type							15	1
Northern blot								
OXD literature index(2)								
cDNA source data(40)								

Other database links:

- EC: 1.17.99.3
- DotS: [OT.527949](#), [OT.94351156](#)
- UniGene: 28720
- TIGR: [TC.1557175](#)
- NIA Mouse Gene Index: [1033228](#)
- Entrez Gene: 92732

Protein domains:

- InterPro ID Description
- IPR002655 Acyl-CoA oxidase, C-terminal
- IPR006091 Acyl-CoA dehydrogenase/oxidase, central region
- IPR002075 Acyl-CoA dehydrogenase/oxidase C-terminal
- IPR009100 Acyl-CoA dehydrogenase/oxidase, middle and N-terminal
- IPR012259 Acyl-CoA oxidase
- IPR013764 Acyl-CoA dehydrogenase, type1/2, C-terminal

Molecular reagents: All nucleic(1) cDNA(41)

References: (Earliest) 167589 Hughes S et al., "Prenatal and postnatal development of peroxisomal lipid-metabolizing pathways in the mouse." *Biochem J* 2001 Feb 1;353(Pt 3):673-80 (Latest) 129580 The FANTOM Consortium and RIKEN Genome Exploration Research Group and Genome Science Group (Genome Network Project Core Group), "The Transcriptional Architecture of the Mammalian Genome." *Science* 2005; 309(5740):1569-1563

Exercise 3; Step 5a results for gene symbol link to Acox2

SNP Detail

ID/Version: rs30597997 dbSNP
 Additional resources: MGI | MGI Mouse Gbrowse | Ensembl SNPView | UCSC Browser | NCBI MapViewer
 Created in dbSNP build: 125
 Last updated in dbSNP build: 125

Variation: Type: SNP
 Alleles: A/T

Genome location and flanking sequence: Location: Chr14:7043734 (NCBI Build 36)
 SNP orientation to the genome: forward

SNP reference flanking sequence:
 TACTTGGAT TGTGTATCTG TGTCTCCAC CCTTGCATTA CCTCTAACAT TTTTGGAACT
 CTCTAGGGGT CAGGGGCGAC TGAGTCTCCA TATTATGGC CATTTTCTCT GATGCTCTGG
 ACATTGGAG ACTAGAGGTT TATGGAGCC CAGCTTCAAT AATCTCCCA CAGCTGGAC
 AGCATTGCC ACCCTCTATA AGCAGTCCC AGGAAGCAGA GTGTACTCCA GGCAGGAGCC
 TGAAGTCTT CTTCAGATA TCACTTAGG AGCTGTGAA GAATTOTAA AGGCTGGTGG
 W
 CATGAAATGC AAGGCATGC TCACAGCAAG CTGAGGCGAG AGTTTCTGCT GCTGTGTCTG
 GTATTCCAGG ATTTTGGCT CTGGGTCACT GAGCAGAGAA AAGAAGCACA GOTTTAGGAG
 TTCACTTTTA CACTGTCAAG CCATACAAAT GGACCACTTA AAGGCTGAAA TGAAGCTACT
 GTTTTAA CTTACTAC CACGATATC CAGCATATT TTTTGTATA AATGTGACA
 GCATACAA CCATTATCTT AGCTAAAGT CTGAAAGGTT CTGAACTCA AACCATCTGG
 W
 A/T

Note: Sequence in lower case indicates low-complexity or repetitive sequence

BLAST SNP flanking sequence against the mouse Genome
 send flank to MouseBLAST [Go]

SNP assays and SNP consensus: Strain alleles of submitted assays, and consensus values for this SNP

Assay ID	Submitter SNP ID	Submitter Handle	Population	SS orient	Variation Type	12931/2014	N/J	12781/6/1	BBV/2/1
ss39202423	mCV23258065	ABI	CRAMUS_MOUSE	f	SNP	A	T	T	T

SNP Consensus Information

SNP	Consensus Type	12931/2014	N/J	12781/6/1	BBV/2/1
rs30597997	SNP	A	T	T	T

Gene Marker associations and Function Classes: MGI gene/markers associated with the SNP
 Location: Chr14:7043734 (NCBI Build 36) rs-orient: forward

Symbol	Name	Transcript	Protein	Function Class	Allele	Residue	Codes Position	AA Position
Acox2	acyl-Coenzyme A oxidase 2, branched chain	NM_053115 MGI Seq Detail	NP_444345 MGI Seq Detail	Coding-NonSynonymous	T	S	1	360
		NM_053115 MGI Seq Detail	NP_444345 MGI Seq Detail	Contig-Reference	A	T	1	360

Exercise 3; Step 5b results for MGI SNP details for Acox2

Exercise 4: SNP summary information in MGI

- Go to the MGI home page
 - <http://www.informatics.jax.org>
- Go to the **Strains and Polymorphisms** query form
- Instead of selecting **SNPs**, click on the link to the *Summary Report of SNP Data in MGI*
 - The summary report provides information on the number of SNPs per strain per chromosome as well as a tally of how much SNP data are in dbSNP from different providers.

Additional Hints and Tips about using the MGI Mouse SNP Query Form:

- You don't have to limit yourself to looking for SNPs within genes. In the Associated genes section of the SNP query form you can select an option to look for SNPs up to 1000 kb upstream and/or downstream of a target gene
- Using Output format selection box in the Output Options section of the SNP query form you can select to have your query results saved in one of several formats, including as a tab-delimited file.
- By default the SNP query form limits the number of results to 1000. But you can over-ride this default by changing the number in the Output Options section of the SNP query form.
- By default, your query results omit SNPs that map to multiple places in the genome. But you can select the Multiple Locations option in the Output Options section of the SNP query form to include these SNPs in your query results.

5. If you only want SNP calls where the values of the Selected Strains are the Same or Different relative to the Reference Strain you can change the value in the SNPs Returned selector which is just underneath the Reference Strain list box on the SNP query form.

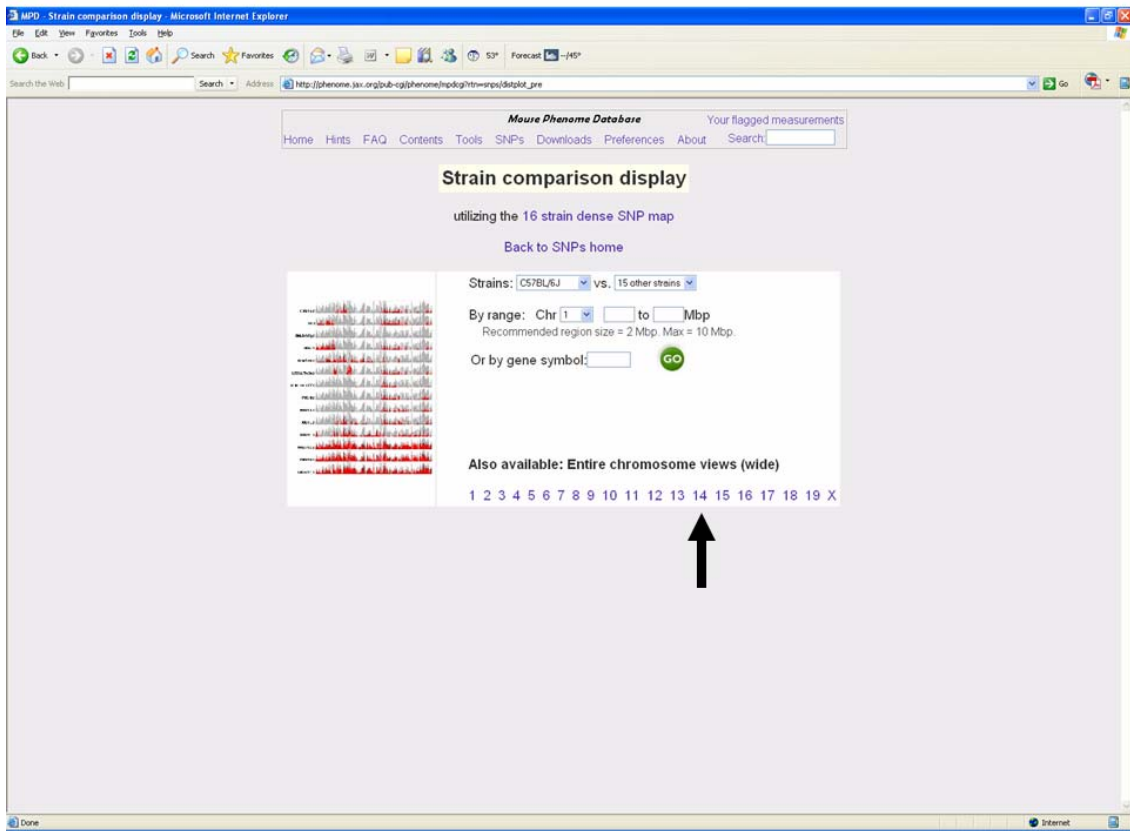
NOTES:

Exercise 5: Strain comparison of SNP data using the Mouse Phenome Database

1. Go to the MPD home page
 - a. <http://www.jax.org/phenome>
2. Click on the link to **Mouse SNPs**
3. Click on the link to Graphical SNP strain comparisons
4. Select one of the chromosome links under Entire chromosome views (wide)
5. The resulting display is a chromosome wide view of SNP density of C57BL/6J relative to 15 other strains. You can click in a region of the display to zoom in.
 - a. At the bottom of the SNP display is a track that shows annotated genes along each chromosome.

The screenshot shows the Mouse Phenome Database website in a Microsoft Internet Explorer browser window. The page title is "Mouse SNPs - Welcome". At the top, there is a navigation menu with links: Home, Hints, FAQ, Contents, Tools, SNPs, Downloads, Preferences, About, and a search box. Below the navigation menu, the main heading is "Mouse SNPs - Welcome". Underneath, there is a section titled "Select one of the following approaches:" with a yellow background. A black arrow points to the first option: "Graphical SNP strain comparison for specified genomic regions up to 10 Mbp, or for entire chromosomes. Uses the 16 strain dense SNP map." The other options are: "Get a listing of SNPs for a gene or region.", "Look up individual SNPs", "View SNPs along with phenotype strain means.", "Download all SNP data for the 20 most-assayed strains in a big zip file, and process locally using your own tools (but not Excel—file is too large).", and "Get more information about this resource, data sources, preparation methods, etc." The browser's address bar shows the URL: http://phenome.jax.org/bub-cg/phenome/npdog/str-wsnp/door.

Exercise 5; Step 3



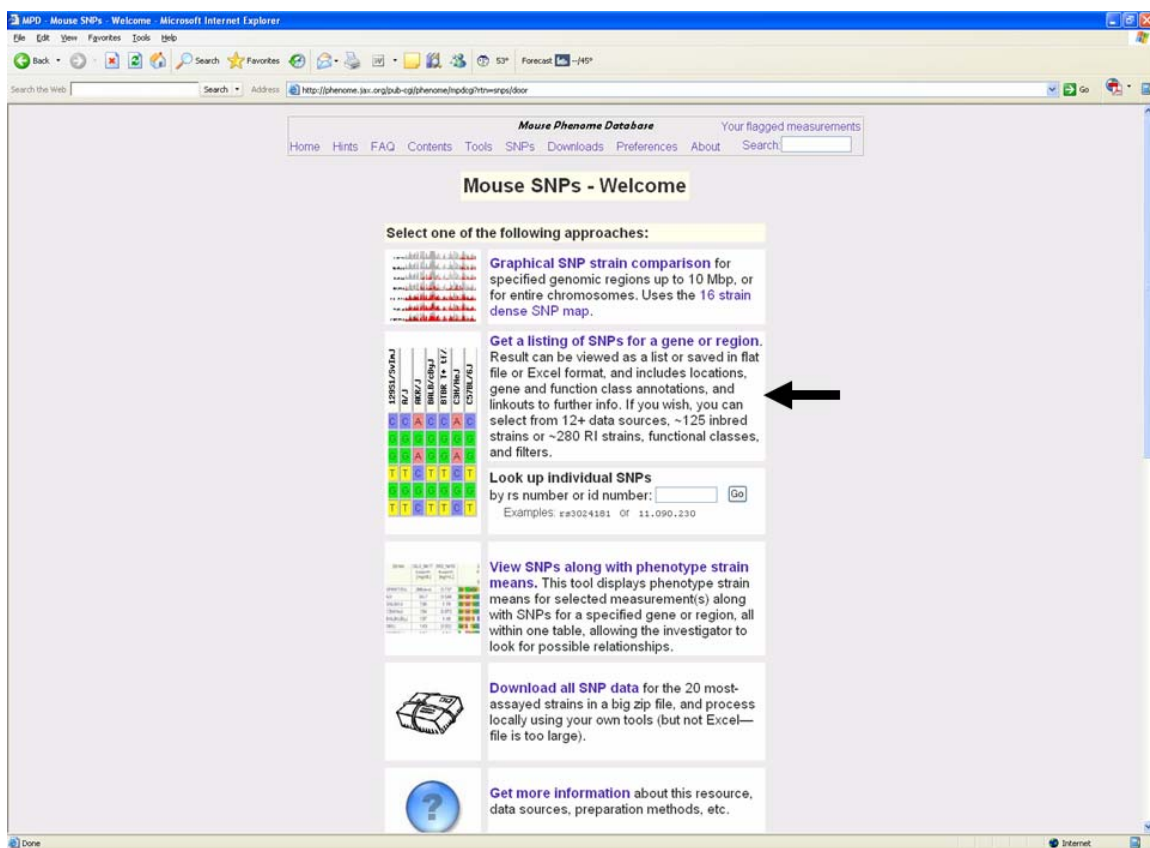
Exercise 5; Step 4



Exercise 5; Step 4 results

Exercise 6: Accessing SNPs from a specific source in a gene or genomic region

1. Go to the MPD home page
 - a. <http://www.jax.org/phenome>
2. Click on the link to **Mouse SNPs**
3. Click on the link to *Get a listing of SNPs for a gene or region*
4. Click on the GO button next to the *Full-flexibility wizard interface*
5. A list of all available SNP data sources is provided along with descriptions of those resources
 - a. This interface allows you to select specific data sources to use to generate a list of SNPs in a gene or region
6. Click on *Unselect All* and wait for the page to refresh
7. Select the Broad1 SNP data source and then click the **Next** button
8. Select the strains you want to include in your query and click the **Next** button
9. Select any filtering options you want and then click the **Next** button
10. The query results show NCBI and Ensembl gene annotations for SNP data side by side for comparison.



Exercise 6; Step 3

Mouse Phenome Database Your flagged measurements

Home Hints FAQ Contents Tools SNPs Downloads Preferences About Search

SNP listing choices

[Back to SNPs home](#)

Result can be viewed as a list or saved in flat file or Excel format, and includes locations, gene and function class annotations, and linkouts to further info. In all cases queries can be refined to add filtering, change strain selection, etc.

- List SNPs within a gene region**
 Gene symbol or description keyword:

(Click here to browse genes)

 - Includes all available data sources.
 - Most-assayed strains
- List SNPs within a specified Mbp region**
 Chr from Mbp to Mbp
 - Includes all available data sources.
 - Most-assayed strains
 - Regions up to 5 Mbp in size.
- Full-flexibility wizard interface**
 - Select from 12+ available SNP data sources
 - Select from ~125 inbred strains or ~280 RI strains
 - Perform polymorphism or other filtering
 - Filter by gene feature / function

Exercise 6; Step 4

Mouse Phenome Database Your flagged measurements

Home Hints FAQ Contents Tools SNPs Downloads Preferences About Search

Mouse SNPs - select source data sets

[Back to MPD SNPs home page](#)

Select from the available SNP source data sets
 Source data sets vary widely in size, number of strains, data density, and uniformity of coverage of genome and strains. Check the boxes next to the data sets most appropriate for your application.

[Click here to select ALL inbred strain data sets](#)

Select data set(s) for inclusion	Year ^[1]	# SNP records ^[2]	Mouse genome coverage click to see graph	# strains	Strain coverage notes click to see list	Mean data density ^[3]	%Polymorphism matrix ^[4]
<input checked="" type="checkbox"/> Broad1	2006	138608	uniform	49 strains	uniform	92 %	yes
<input type="checkbox"/> Celera2	2006	2093327	uniform	5 strains	fairly uniform	73 %	yes
<input type="checkbox"/> Perlegen2	2005	8273434	uniform	16 strains	uniform	89 %	yes
<input type="checkbox"/> TJL3	2005	1994	uniform	107 strains	uniform	92 %	yes
<input type="checkbox"/> WTCHG1	2005	11571	uniform	67 strains	uniform	100 %	yes
<input type="checkbox"/> WTCHG-RI 2005 11571 <input type="checkbox"/> AXB <input type="checkbox"/> BXA <input type="checkbox"/> CXB <input type="checkbox"/> BXD <input type="checkbox"/> BXH <input type="checkbox"/> AKXD <input type="checkbox"/> LXS <input type="checkbox"/> SWXJ			uniform	varies	uniform	~100%	[4]
<input type="checkbox"/> GNF2	2004	9594	uniform	48 strains	uniform	89 %	yes
<input type="checkbox"/> Roche2	2004	69781	fairly uniform with major concentrations	18 strains	fairly uniform	78 %	yes
<input type="checkbox"/> TJL2	2004	228	fairly uniform	48 strains	uniform	84 %	yes
<input type="checkbox"/> GNF1	2003	10790	fairly uniform with some concentrations	9 strains	fairly uniform	83 %	yes
<input type="checkbox"/> TJL1	2003	28	1 or 2 per chromosome	96 strains	uniform	99 %	yes
<input type="checkbox"/> WICGR3	2003	261521	uniform	5 strains	predominantly 2 strain alleles per location, usually C57BL/6J vs. CZECHII/EIJ	40 %	yes
.....	fairly uniform with some	predominantly 2 strain alleles per location.

Exercise 6; Step 5, 6, 7

MPD - Mouse SNPs - select strains - Microsoft Internet Explorer

Address: http://phenome.jax.org/sub-cgi/phenome/mpdog?trn=srps%2fchoosestrainslip_Broad1=on

Mouse Phenome Database

Home Hints FAQ Contents Tools SNPs Downloads Preferences About Search

Mouse SNPs - select strains

MPD SNPs home page Back to data sets

Select strains of interest. All strains from the **Broad1** data set are listed with total number of SNPs available. Strains are listed in alphabetical order (click here to rank by #SNPs). [Next](#)

Your recent selection
Select all

Strain	#SNPs *	..continued	#SNPs *	..continued	#SNPs *
<input checked="" type="checkbox"/> 129S1/SvImJ	131375	<input type="checkbox"/> CZECHII/EiJ	122699	<input type="checkbox"/> NZW/LacJ	126868
<input type="checkbox"/> 129S4/SvJae	128917	<input type="checkbox"/> DBA/1J	130441	<input type="checkbox"/> O20	125947
<input type="checkbox"/> 129X1/SvJ	132540	<input type="checkbox"/> DBA/2J	130359	<input type="checkbox"/> PERA/EiJ	124030
<input type="checkbox"/> A/J	133909	<input type="checkbox"/> DDK/Pas	128517	<input type="checkbox"/> PL/J	130818
<input type="checkbox"/> AKR/J	127773	<input type="checkbox"/> FVB/NJ	125981	<input type="checkbox"/> PWD/PhJ	119183
<input type="checkbox"/> BALB/cByJ	126862	<input type="checkbox"/> I/LnJ	129021	<input type="checkbox"/> Osi5	127947
<input type="checkbox"/> BTBR T+ tf/J	128501	<input type="checkbox"/> JF1/Ms	121320	<input type="checkbox"/> RIIS/J	129694
<input type="checkbox"/> BUB/BnJ	130585	<input type="checkbox"/> KK/HIJ	129310	<input type="checkbox"/> SEA/GnJ	129089
<input type="checkbox"/> C3H/HeJ	130363	<input type="checkbox"/> LG/J	128871	<input type="checkbox"/> SEG/Pas	105393
<input checked="" type="checkbox"/> C57BL/6J	138608	<input type="checkbox"/> LP/J	129994	<input type="checkbox"/> SJL/J	129433
<input type="checkbox"/> C57BLKS/J	131486	<input type="checkbox"/> MAMyJ	129063	<input type="checkbox"/> SM/J	118672
<input type="checkbox"/> C57BR/cdJ	130396	<input type="checkbox"/> MAI/Pas	120810	<input type="checkbox"/> SPRET/EiJ	106489
<input type="checkbox"/> C57LJ	128964	<input type="checkbox"/> MOLF/EiJ	119187	<input type="checkbox"/> ST/lJ	129301
<input type="checkbox"/> C58/J	131782	<input type="checkbox"/> MSMMs	121763	<input type="checkbox"/> SWR/J	129821
<input type="checkbox"/> CAST/EiJ	119403	<input type="checkbox"/> NOD/LtJ	128088	<input type="checkbox"/> WSB/EiJ	130128
<input type="checkbox"/> CBA/J	130051	<input type="checkbox"/> NON/LtJ	125238		
<input type="checkbox"/> CE/J	127576	<input type="checkbox"/> NZB/BINJ	127115		

Exercise 6; Step 8

MPD - Mouse SNPs - settings - Microsoft Internet Explorer

Address: http://phenome.jax.org/sub-cgi/phenome/mpdog?trn=srps%2fsettings?proj=Broad1&pmapped=yes&S3=on&S7=on

Mouse Phenome Database

Home Hints FAQ Contents Tools SNPs Downloads Preferences About Search

Mouse SNPs - settings

New query/ SNPs home Select data sets Select strains

You're currently working with the Broad1 data set

Select a filtering mode

- No filtering (but rows must have at least 2 strain alleles present)
- SNP locations must have **polymorphic alleles** for certain strains (to be selected next...)
- SNP locations must have **same alleles** for certain strains (to be selected next...)
- SNP locations must have **all alleles present** for the strains selected below

Result will be limited to SNP rows that have an allele present (nucleotide, insertion, deletion) for each of the selected strains.

Display only the strains selected here

Select genomic region

- By location range: Chr= 1 From 10,000 Mbp To 15,000 Mbp
- By gene symbol (browse)
- All of chromosome 1
- Entire genome

Retrievals are limited to 10 Mbp where Perlegen2 or Celera2 are involved. Any retrieval exceeding 30 seconds of cputime will be interrupted and incomplete. Please do

Exercise 6; Step 9

MPD - View / download mouse SNPs - result - Microsoft Internet Explorer

Mouse Phenome Database

View / download mouse SNPs - result

11481 result rows. Query summary %Polymorphism matrix

Download (zip file) Help RS list New query / Refine

Showing the first 200 rows...

Mbp location	NCBI gene annotation (Help)	Ensembl gene annotation (Help)	dbSNP SNP annotation (Help)	dbSNP rs	dbSNP observed	Source	Insertion alleles
1 3.013441				T A		Broad1 1-3027623 1	
1 3.036178				C A		Broad1 1-3050112 1	
1 3.036265				G A		Broad1 1-3050199 1	
1 3.051362				G A		Broad1 1-3065296 1	
1 3.051854				G A		Broad1 1-3065788 1	
1 3.062749				A C		Broad1 1-3076683 1	
1 3.063538				G C		Broad1 1-3077472 1	
1 3.093816				T C		Broad1 1-3107750 1	
1 3.093903				T A		Broad1 1-3107837 1	
1 3.095984				A C		Broad1 1-3109918 1	
1 3.108194				A C		Broad1 1-3122128 1	
1 3.119110				G A		Broad1 1-3133044 1	
1 3.119136				T C		Broad1 1-3133070 1	
1 3.125333				C C		Broad1 1-3139267 1	
1 3.125499				G A		Broad1 1-3139433 1	
1 3.133148				G T		Broad1 1-3147082 1	
1 3.156821				T C		Broad1 1-3170755 1	
1 3.182468				G A		Broad1 1-3196402 1	

Exercise 6; Step 10

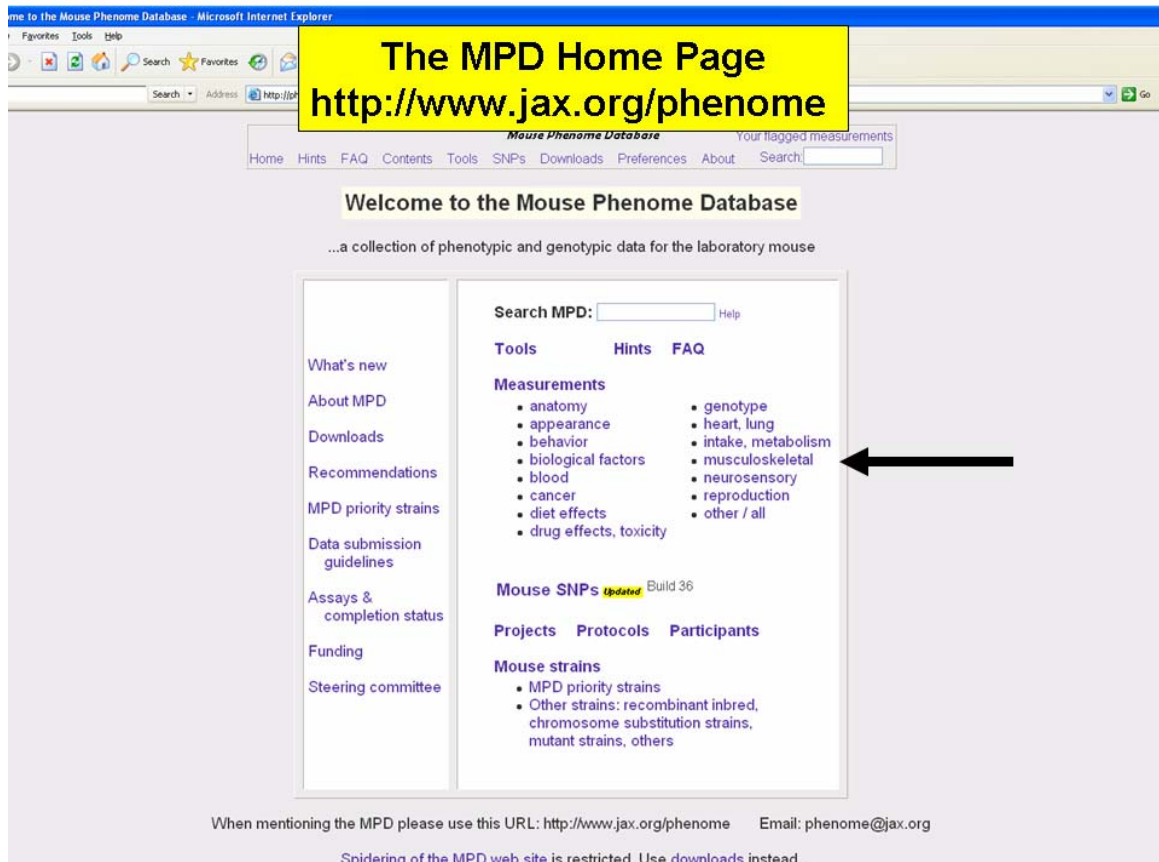
Exercise 7: Integrating SNPs and phenotypic measurement data in MPD

In order to associate SNP data with phenotypic measurement data in MPD you must first select the measurement data that you are interested in. In this Exercise you will select data from a musculoskeletal project that was submitted to MPD. However MPD also allows you to create your own custom set of measurements from many different projects. See the MPD FAQ page for additional details on how to flag measurements and create your own custom data sets (<http://phenome.jax.org/pub-cgi/phenome/mpdcgi?rtn=docs/aboutmpd>).

MPD has many additional data analysis tools other than the SNP analysis tools demonstrated in this tutorial!

1. Go to the MPD home page
 - a. <http://www.jax.org/phenome>
2. Click on the Measurement category of “musculoskeletal”
3. Click on the specific measurement category of bone mineral density and content
4. Three different projects are in MPD with data related to bone mineral density. Select the Naggert1 project.
5. From the Naggert1 Project page, select the “Other tools” icon

6. Click on the “Strain means w/genes & SNPs” tool icon
7. On the next page, all of the measurements for the Naggert1 project are listed. Select GLU_foldchng_fat17 and then click on the GO button.
8. Sort the rows by GLU_foldchng_fat17
9. Change display to Z-magnitude bars
10. Enter a chromosomal region or a gene name
11. Click on the **Go** button
12. SNPs in the genes or regions of interest are displayed along side the phenotype measurement data.



Exercise 7; Step 2

MPD - Measurement categories - Microsoft Internet Explorer

Home Hints FAQ Contents Tools SNPs Downloads Preferences About Search

Measurement categories

List all MPD measurements by category List all by project List all in Excel by category .by project

Top-level categories:

- anatomy
- appearance
- behavior
- biological factors
- blood
- cancer
- diet effects
- drug effects, toxicity
- genotype
- heart, lung
- intake, metabolism
- musculoskeletal**
- new category
- reproduction
- other / all

Browse measurement categories:

- acoustic startle response
- activity and motor function
- adrenal, thymus, kidney
- anxiety
- atherogenic diet
- avoidance
- blood calcium and pH
- blood coagulation factors
- body composition
- body weight
- bone mineral density and content**
- brain measurements
- cardiovascular
- cholesterol
- coat color and appearance
- craniofacial measurements**
- gallbladder and gallstones
- glucose
- hearing
- hematology
- hormones
- lipids
- lipoproteins
- liver
- lungs
- metabolism
- metastatic progression
- MHC H2 haplotype
- nociception
- organ weights
- pathology images
- peptides and proteins
- prepulse inhibition

Exercise 7; Step 3

MPD - MPD category: bone mineral density and content - Microsoft Internet Explorer

Home Hints FAQ Contents Tools SNPs Downloads Preferences About Search

MPD category: bone mineral density and content

Available subcategories:

- body
- body, atherogenic diet
- femur
- spine

Projects supplying data:

- Jaxwest1 (7 strains)
- Naggert1 (43 strains)**
- Tordoff3 (40 strains)

Tools control panel Examine a strain Exceptional strains Compare 2 strains All MPD categories

bone mineral density and content
Units & abbreviations
Click on a measurement description for summary, and to compare strains.

	Short name	Actions
body		
bone mineral content [g] Tordoff3 MPD 10327	BMC	🔍 🗑️
bone mineral content (BMC) [g] Jaxwest1 MPD 15131	BMC	🔍 🗑️
bone mineral density [g/cm ³] Tordoff3 MPD 10326	BMD	🔍 🗑️
bone mineral density (BMD) [g/cm ³] Jaxwest1 MPD 15130	BMD	🔍 🗑️
body, atherogenic diet		
bone mineral content (BMC) after 8 wks on atherogenic diet [g] Naggert1 MPD 14314	BMC_fat8	🔍 🗑️
bone mineral density (BMD) after 8 wks on atherogenic diet [g/cm ³] Naggert1 MPD 14312	BMD_fat8	🔍 🗑️
femur		
bone mineral content (BMC) of right femur [g] Jaxwest1 MPD 15133	BMC_femurR	🔍 🗑️
bone mineral density (BMD) of right femur [g/cm ³]		🔍 🗑️

Exercise 7; Step 4

MPD - Project: Naggert1 - Microsoft Internet Explorer

Address: http://phenome.jax.org/pub/cgi/phenome/tpd/cgi-bin=projects/details?m=Naggert1

Mouse Phenome Database

Project: Naggert1

Naggert JK, Svenson KL, Smith RV, Paigen B, Peters LL - Diet effects on bone mineral density and content, body composition, and plasma glucose, leptin, and insulin levels
MPD 143 Naggert1 (2003)

- Participants, publications, funding
- Naggert1 measurements
- Naggert1 protocol
- Naggert1 animal documentation
- Strains and animal notes
- Notes pertaining to the data set
- Downloadable project files

Strain comparison plots | Scatterplots | Other tools

- List MPD-wide correlations
- Examine one strain
- Exceptional strains
- Compare 2 strains
- Numbers of mice tested

Naggert1 measurements

Key to units & abbreviations
Note: data collection is ongoing; more data expected
Click on a measurement description for summary, and to compare strains

Measurement	Short name	Actions
glucose, before beginning atherogenic diet [mg/dL] MPD 14302	GLU	[View] [Compare]
atherogenic diet		
glucose, after 17 wks on atherogenic diet [mg/dL] MPD 14303	GLU_fat17	[View] [Compare]
fold change in glucose after 17 wks on atherogenic diet [fold] MPD 14304	GLU_foldchg_fat17	[View] [Compare]

hormones

Exercise 7; Step 5

Cannot find server - Microsoft Internet Explorer

Address: http://phenome.jax.org/pub/cgi/phenome/tpd/cgi-bin=views/tools?regression=Naggert1

Mouse Phenome Database

Select a tool by clicking on its graphical icon. (context: Naggert1)
(Hover mouse pointer over graphical icons to see popup short text descriptions of tools)

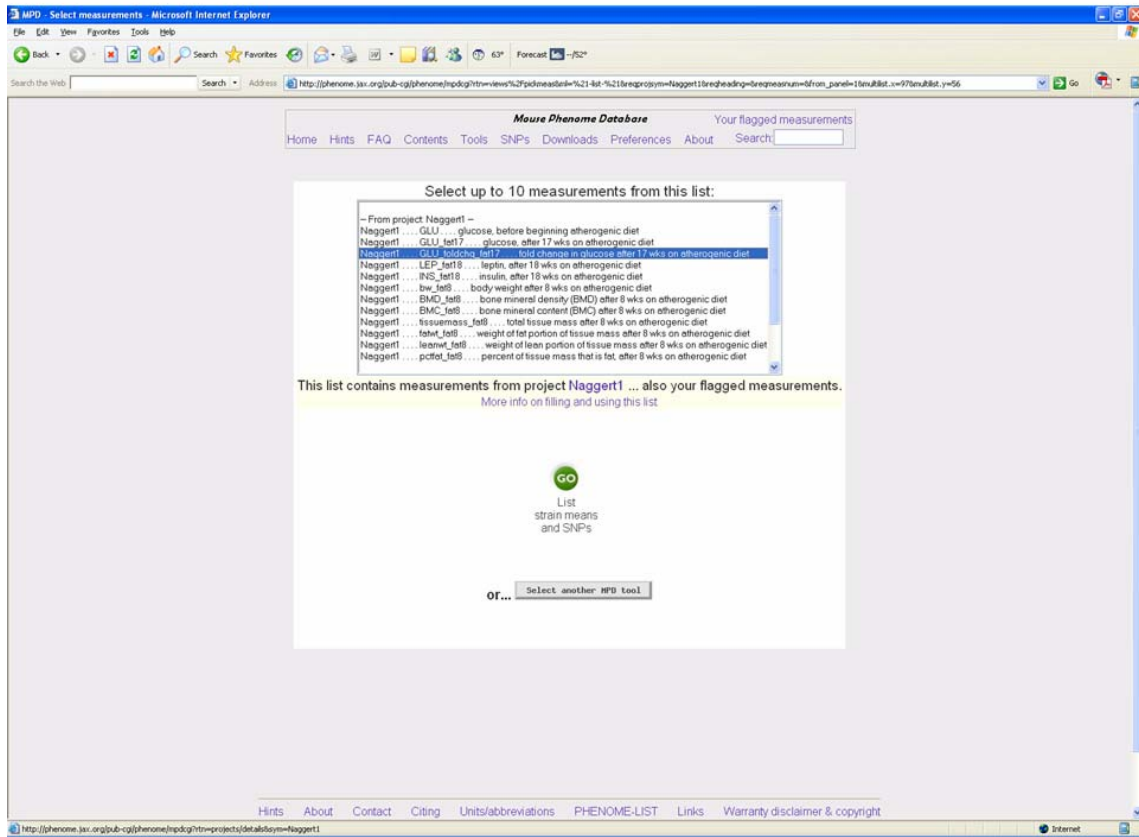
Strain comparison plots (More info)	Scatterplot	Correlations matrix or list	Scatterplot matrix
Find mouse models	Sex differences	Ratios & differences for 2 measurements	Strain coverage table
Strain means w/ genes & SNPs	Graphical strain pair comparisons	List differences for two strains	Download customized set of pheno data
			SNP tools

Also available:

For the Naggert1 project:

- Download the entire Naggert1 project data set
- Naggert1 correlations across all MPD
- List Naggert1 values for a particular strain
- List exceptional strains in Naggert1
- Rank Naggert1 measurements by # flaggings

Exercise 7; Step 6



Exercise 7; Step 7

MPD - Table of strain means for several measurements, with genes & SNPs - Microsoft Internet Explorer

Select other measurements / other MPD tools

Sort the rows by: **GLU_foldchg_fat17 (Naggert1)** Use reverse order Z-score magnitude bars

Show SNPs* for: **Slc2a1** in region

Enter one or more gene symbols, rs#, ranges. Click here for other SNP tools

See explanatory notes at bottom of page.

Female			Male		
Strain	GLU_foldchg_fat17 Naggert1 [fold] Sorted	SNPs* in gene Slc2a1	Strain	GLU_foldchg_fat17 Naggert1 [fold] Sorted	SNPs* in gene Slc2a1
WSB/EIJ	2.74 (N=4)	...	NOD/LJ	2.74 (N=4)	...
NOD/LJ	BTBR T+ tf/J
BTBR T+ tf/J	NZW/LacJ
FVB/NJ	DBA/2J
DBA/2J	C57BL/6J
NZW/LacJ	C3H/HeJ
C3H/HeJ	CAST/EIJ
C57BL/6J	129S1/SvImJ
KK/HJ	WSB/EIJ
CAST/EIJ	FVB/NJ
AKR/J	MOLF/EIJ
129S1/SvImJ	A/J
MOLF/EIJ	AKR/J
A/J	KK/HJ

More options: Warn when N mice < 5 SNPs must have 4+ eligible strain alleles to be included.
If SNP columns are present, omit strain rows that have < 6 SNP alleles [Go]

Exercise 7; Steps 8,9,10,11

Bonus Exercise:

The Heart, Lung, Blood, Sleep disorders center at The Jackson Laboratory has mapped a recessive neutrophil mutant, hlb156 (MGI:2678700) to Chromosome 17 between genetic markers D17Mit133 and D17Mit175. The mapping cross included C57BL/6J and 129S1/SvImJ.

1. Go to the MGI home page
 - a. <http://www.informatics.jax.org>
2. Click on the link to the **Strains and Polymorphisms** query forms
3. Click on the link to the **SNPs** query form
4. Select *C57BL/6J* as the reference strain and then select *129S1/SvImJ* as a selected strain.
5. Enter chromosome 17 and marker symbols D17Mit133 and D17Mit175 in the Marker Range text box.
 - a. Note that when you search for a specific gene or marker range that the SNP query results have additional links to MGI resources such as a list of the genes and a link to the Mouse GBrowse interactive sequence feature map.
6. Click on Search and explore the query results.
7. Try refining your searches by SNP type and SNP function class.

End of Tutorial.