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Exercises for Mouse Genome Informatics (MGI) (Version 3, 2008)

You can choose to do these using the questions as your only guide—or see the following pages for the step-by-step checklist to finding these answers.

1. Find receptor genes on mouse chromosome 1 which have been characterized as having a role in the diabetes phenotype. Examine homologous genes in other mammalian species. Find a protein sequence for this gene and obtain the FASTA format output.

Skills: Genes and Markers search with phenotypes, mammalian homology, obtaining protein sequences

2. Can you find genes in the Patched family, which have SNPs in the coding region, and examine a selected SNP for its occurrence in many strains?

Skills: Strains and Polymorphisms search, SNP function class selection, SNP details pages

3. Use the Gene Expression Data query to identify genes involved in Notch binding, which have been detected in brain by in situ hybridization. Find mutant mice which have been examined in this manner, and view the expression data images.

Skills: Gene expression data query, output option changes, image viewing

Mouse Genome Informatics Exercises, version 3. Correspond to the data available in December 2008.

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Step-by-Step checklist/instructions for MGI exercises

1. Find receptor genes on mouse chromosome 1 which have been characterized as having a role in the diabetes phenotype. Examine homologous genes in other mammalian species. Find a protein sequence for this gene and obtain the FASTA format output.

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Step	Action	V
1	From the MGI HomePage (<u>http://www.informatics.jax.org/</u>), click	
	on the top menu labeled "Search".	
2	From the Search menu, click on the "Genes & Markers Query"	
	item in the Genes group.	
3	On the Genes and Markers Query Form page, you will make	
	several selections for search options. First, type "receptor" in the	
	Genes/Marker Symbol/Name text box.	
4	Select chromosome 1 in the Map Position area.	
5	Type "diabetes" in the Mouse phenotypes and mouse models of	
	human disease area.	
6	Click the Search button to run the query.	
7	From the results page, click the link for Irs1, the insulin receptor	
	substrate 1 gene.	
8	Examine the Irs1 page. In the Mammalian homology section,	
	click the link for Mammalian Orthology. You should see a new	
	page with information about related genes in several species.	
9	Use the Back button to return to the Irs1 page.	
10	In the Sequences section, click the link for All Sequences (23).	
	Note: the number of sequences may change over time.	
11	On the Mouse Sequences Summary Report, note that you can	
	access DNA, RNA and protein (polypeptide) sequences. Scroll	
	through the list of sequences and click the checkbox next to the	
	first polypeptide in the list.	
12	At the bottom of the page, ensure that the pulldown menu	
	selection says "download in FASTA format".	
13	Click the Go button beneath the menu.	
14	A new page will present the amino acid sequence for the selected	
	protein. You could copy and paste this sequence into many	
	analysis tools.	
15	Use the back button to return to the Irs1 page. Click the MGI	
	logo at the top left to return to the MGI homepage.	



2) Can you find genes in the Patched family, which have SNPs in the coding region, and examine a selected SNP for its occurrence in many strains?

Step	Action	V
1	From the MGI homepage http://www.informatics.jax.org/, use the	
	Search pulldown menu and highlight the "Strains / SNPs"	
	option.	
2	From the Strains/SNPs menu option, click the SNP Query menu	
	item.	
3	On the Mouse SNP Query Form page, we will make several	
	selections in the next steps. Make sure you are following each one	
4	carefully to obtain the expected results.	
4	In the Strains and strain comparisons section, select these strains	
	Strains boy	
	• A/.1	
	• FVB/NJ	
5	In the SNP attributes section select two options in the dbSNP	
	Function Class box:	
	Coding-NonSynonymous	
	Coding-Synonymous	
6	In the Associated genes area, make these choices:	
	Pulldown menu: begins	
	Text box: patched	
	 Search menu: current symbols/names 	
7	Leave all other settings as default. Click Search to run the	
	query.	
8	A new web page will launch that presents the results. How many	
	genes are displayed?	
	At this time, 4 genes are shown: Ptch1, Ptch2, Ptchd2, Ptchd3.	
9	From the results list, find the first SNP listed which shows data	
	for all 3 strains. (Currently this is rs134/7933 in Ptch2 but that	
	link in the first column	
10	On the SNP detail page, are there other strains that carry either of	
10	these alleles?	
	Yes, at this time 10 strains carry the T allele, and 38 carry the C	
	allele.	
11	Click the link for MGI Home at the top left of the MGI page to	
	return to the homepage.	



3) Use the Gene Expression Data query to identify genes involved in Notch binding, which have been detected in brain by in situ hybridization. Find mutant mice which have been examined in this manner, and view the expression data images.

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Step	Action	V
1	From the MGI homepage http://www.informatics.jax.org/, click the	
	image labeled "Expression" from the "Explore MGI" area.	
2	Click the link for Gene Expression Data Query.	
3	On the Gene Expression Data Query Form you will make several	
	changes. Follow the instructions to obtain the expected outcome.	
4	In the Gene Ontology classification section, enter this text in the	
	text box:	
	 notch binding 	
5	In the Expression area, type "brain" in the Anatomical	
	Structures section.	
6	In the Assay type(s) area, select RNA in situ .	
7	Click Search to run the query.	
8	On the result page, determine how many genes meet these criteria.	
	(At this time there are 4 genes: Dll1, Dll3, Jag1 and Jag2).	
9	From the results in this format it isn't clear where or when the	
	expression is examined. Use the back button to return to the	
	Query Form.	
10	In the Sorting and output format area, change the radio button	
	near Return data type to: Assay Results.	
11	Click Search.	
12	What is the earliest expression stage for DII1?	
	(At this time it is embryonic day 8.0)	
13	Is DII1 detected in Presenilin 1 mutant mice (Psen1)?	
	(Yes, we can see DII1 detected in some Psen1 mutants.)	
14	In the Psen1 mutant mice section (Psen1 ^{tm1Shn} /Psen1 ⁺) click the link:	
	<u>data</u> (MGI:1859916).	
15	On the Gene Expression Data Query Results Details page, click	
	the <u>6Aa</u> link in the column with the camera icon to view the data	
	for:	
	• <u>TS19: telencephalon; ventricular layer</u> Figure <u>6Aa</u>	
16	A new page should open with image data to show the results.	