

JAX Synteny Browser Use Cases

<http://syntenybrowser.jax.org/>

June 2019

The basic workflow for the JAX Synteny Browser involves four steps:

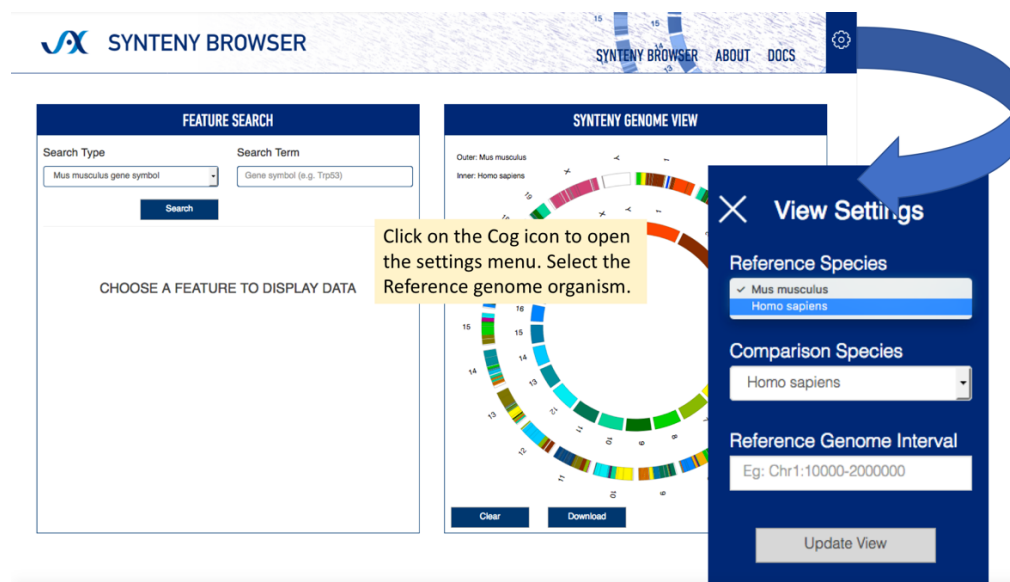
- (1) selecting the Reference genome,
- (2) specifying a region of interest on the Reference,
- (3) visualizing the region of interest and its corresponding conserved syntenic block(s) in the Comparison genome, and
- (4) selectively highlighting genes in the Reference and Comparison genomes based on their biological attributes.

Two specific use cases are described below serve to demonstrate this workflow.

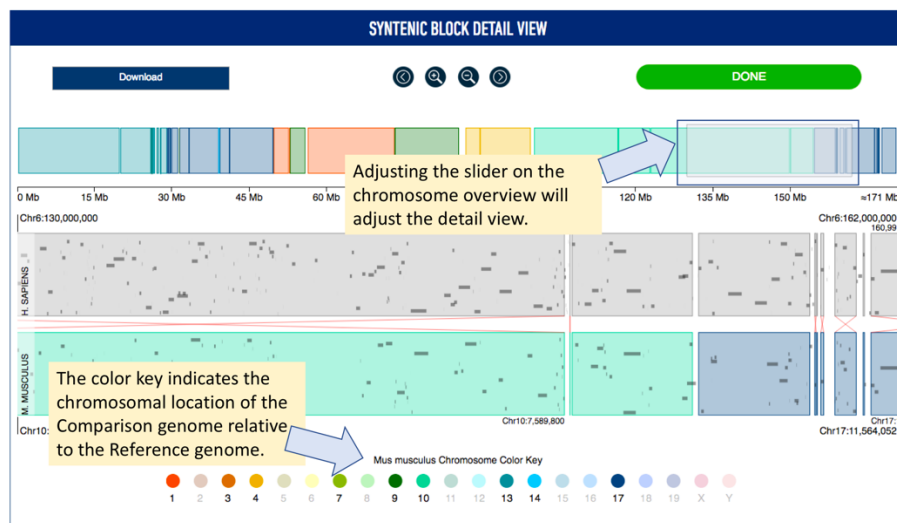
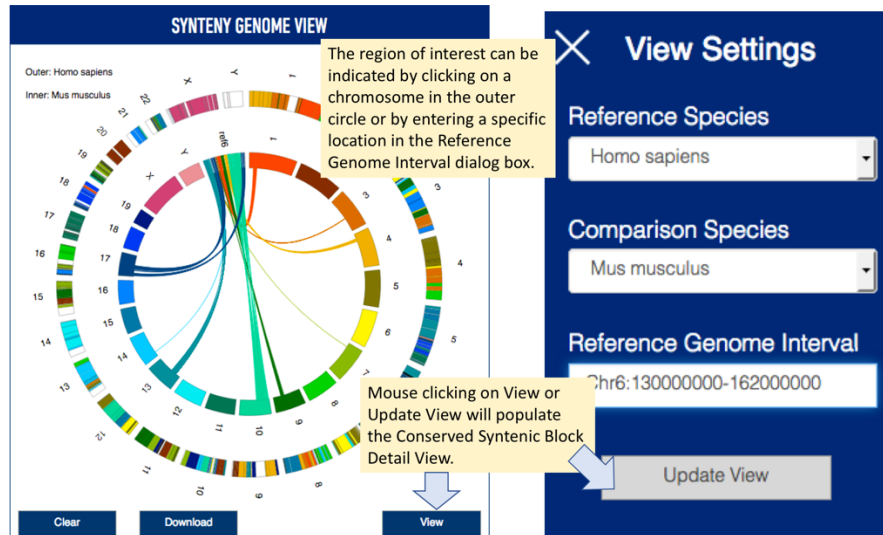
Use Case #1: Identify candidate genes in a mapped interval for human lung cancer susceptibility

A region of human chromosome 6 (6q23–25; GRCm38 chr6: 130300000-161000000 bp) was identified previously as a linkage interval associated with human lung cancer susceptibility (Bailey-Wilson, et al., 2004). Because this linkage interval also overlaps regions of allelic loss observed in several cancers, the authors hypothesized that genes involved in regulating apoptosis would be good candidates for the susceptibility phenotype.

The first step in using the JAX Synteny Browser to find potential candidate genes for the lung cancer susceptibility locus is to select human as the Reference genome using the Settings menu.



Next, the user navigates to the region of interest on chromosome 6 using one of two options. For the first option, human chromosome 6 is selected in the Synteny Genome View graphic. Clicking on the View button in this panel results in the entire chromosome being displayed in the Syntenic Block Detail window. The display interval can then be refined interactively using the slider on the chromosome overview graphic. Alternatively, the coordinates of a genomic interval for the Reference genome can be entered in the appropriate dialog box within the Settings menu. When the “Update View” button is selected, the user-specified genomic region is then displayed in the Syntenic Block Detail panel.



CURRENT FILTER SELECTION

☐ HIDE ALL GENOME FEATURES THAT DONT MATCH FILTER REQUIREMENTS

Filter Criteria 1 (FC1):
Find Gene by ID or Symbol

Filter Criteria 2 (FC2):
Find Gene(s) by Type
☒ lncRNA gene (2)
☐ miRNA gene (2)
☐ polymorphic pseudogene (2)
☒ protein coding gene (996)
☐ pseudogene (6)
☐ IN REFERENCE ☐ IN COMPARISON ☒ IN EITHER

Filter Criteria 3 (FC3):
Find Gene(s) by Ontology

☐ IN REFERENCE ☐ IN COMPARISON ☒ IN EITHER

Selection Options:
JR is default operation)

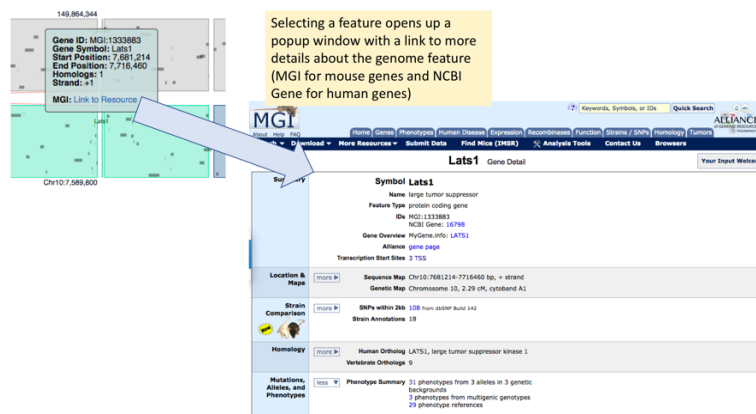
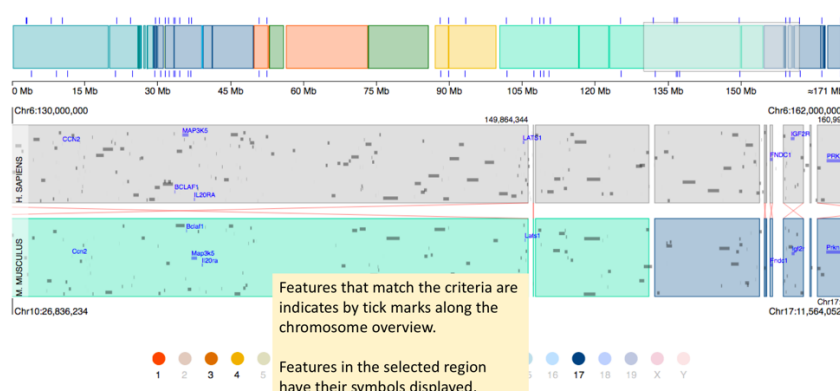
({ FC2 } ☒ AND { FC3 }) ☐ AND ...
☐ OR ☒ OR

DOWNLOAD CSV

Filter:

Showing 1 to 37 of 37 entries

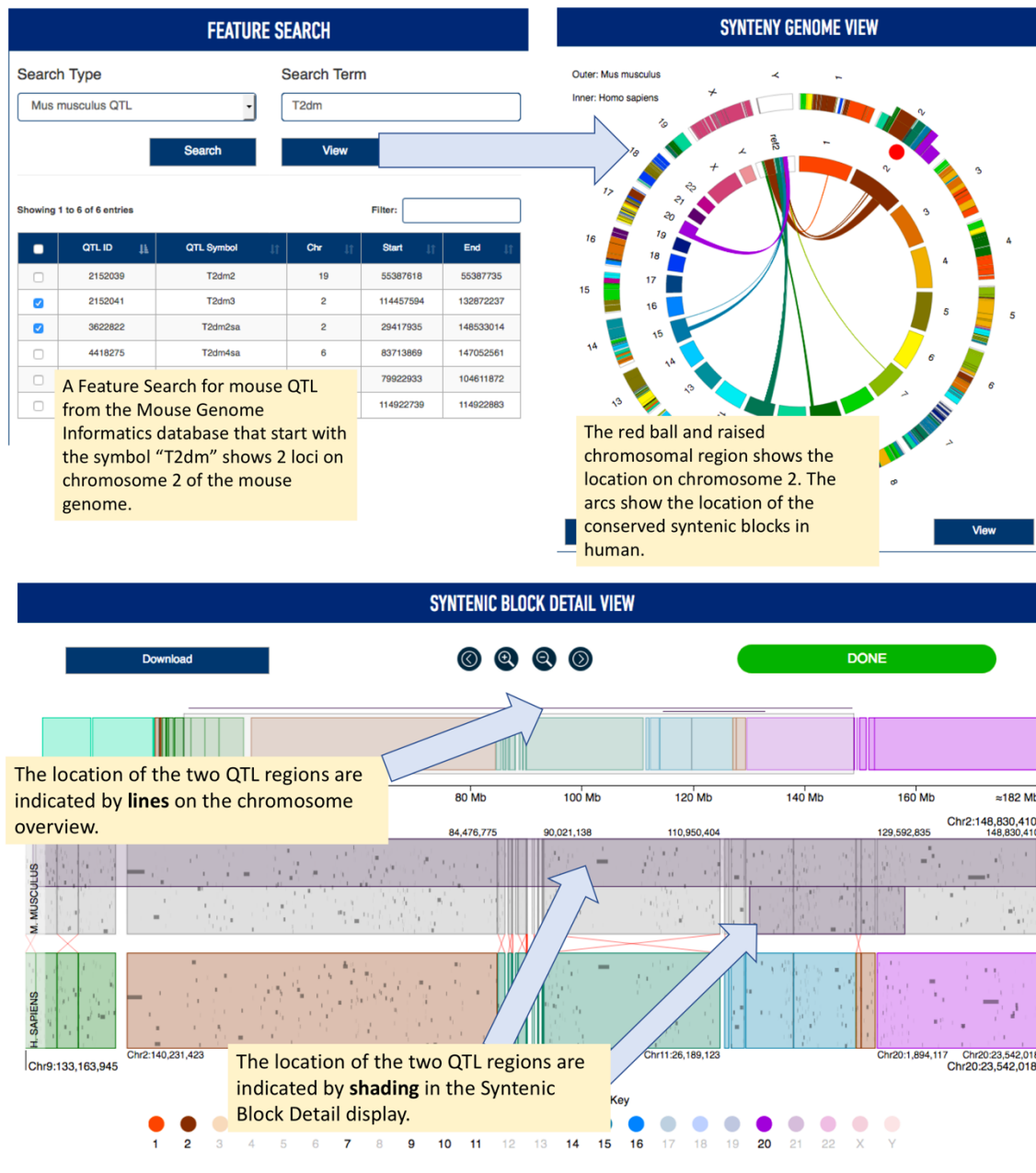
Gene Symbol	Gene ID	Cnr	Start	End	Strand
Ager	MGI:893592	17	34597962	34600936	+1
Bak1	MGI:1097161	17	27019812	27028639	-1
	MGI:1917580	10	20312078	20342644	+1
	MGI:1349399	4	32615470	32653271	+1
	MGI:95537	10	24595442	24596863	+1
	MGI:88323	10	43579169	43584265	+1
	MGI:1925584	10	40348308	40483818	+1
	MGI:104615	4	33924593	33948831	+1
	MGI:1197015	17	33909414	33915590	+1
	MGI:1913393	13	38645691	38659028	-1
	MGI:1919127	1	20951626	20990841	+1
	MGI:192576	4	28813131	28967499	+1
	MGI:192576	17	7738569	7827302	-1
	MGI:190081	10	42181845	42276746	-1
	MGI:106604	13	24943152	24982501	+1
	MGI:95615	10	49004833	49788766	-1
	MGI:96435	17	12682406	12769664	-1
Irf1	MGI:3605069	10	19712570	19760053	+1
Lata1	MGI:1333883	10	7681214	7716460	+1
Lta	MGI:104797	17	35203165	35205351	-1
Mdm2a	MGI:196876	10	19916479	20142791	+1



b. Identifying candidate genes for Type 2 diabetes

The Quantitative Trait Locus (QTL) *T2dm3* (MGI:2152041) was identified as a region of mouse chromosome 2 associated with insulin resistance in genetically obese mice (Stoehr, et al., 2000). This region is overlapped by another QTL for impaired glucose tolerance, *T2dm2sa* (Kobayashi, et al., 2006).

The Feature Search option allows users to search for mouse QTL records in the Mouse Genome Informatics (MGI) database. A search for the symbol “T2dm” (type 2 diabetes mellitus) returns several results, including the two QTL on chromosome 2 referenced above. Selecting both *T2dm3* and *T2dm2sa* from the Feature Search results table and then clicking on the View button locates the features on mouse chromosome 2 in the Synteny Genome View panel. Clicking on chromosome 2 and then View from this panel generates the Syntenic Block Detail panel which shows the relative location of the two QTL regions to each other.



CURRENT FILTER SELECTION

matched results: (25)

☐ HIDE ALL GENOME FEATURES THAT DON'T MATCH FILTER REQUIREMENTS

Filter Criteria 1 (FC1):

Find Gene by ID or Symbol

gene symbol (e.g. Trp53)

Filter Criteria 2 (FC2):

Find Gene(s) by Type

-- no selection --
 antisense lncRNA gene (231)
 gene (1460)
 intronic lncRNA gene (16)
 lncRNA gene (260)

☐ IN REFERENCE
 ☐ IN COMPARISON
 ☒ IN EITHER

Filter Criteria 3 (FC3):

Find Gene(s) by Ontology

Mammalian Phenotype Ontology (MP)

impaired glucose tolerance

☒ IN REFERENCE
 ☐ IN COMPARISON
 ☐ IN EITHER

Selection Options:
(OR is default operation)

(-- ▾)
☒ AND
-- ▾)
☐ AND
-- ▾)

☐ OR ☒ OR

CLEAR
RUN

[DOWNLOAD CSV](#)

Filter:

Showing 1 to 25 of 25 entries

Gene Symbol	Gene ID	Chr	Start	End	Strand
a	MGI:87853	2	154950204	155051012	+
ApoE1	MGI:1336993	2	127008717	127067909	+
Bta5	MGI:1919819	2	69647171	69667571	+
Bmi1	MGI:88174	2	18677018	18686629	-
Cacnb2	MGI:894644	2	14804053	14988811	+
Chgb	MGI:88395	2	132781278	132795072	+
Commd9	MGI:1923751	2	101886262	101901639	+
Dpm2	MGI:1330238	2	32570858	32573571	+
Epcam	MGI:100000000	2	154568892	154568892	-
Foxo1	MGI:100000000	2	156052947	156052947	-
Gata1	MGI:100000000	2	157347506	157347506	-
Hes6	MGI:100000000	2	174346744	174346744	+
Irf1	MGI:100000000	2	126619299	126619299	-
Pfkfb3	MGI:13149862	2	1044426481	104494446	+
Hnf4a	MGI:109128	2	163506809	163572907	+
Lama5	MGI:105382	2	180176373	180225859	-
Lcn2	MGI:96757	2	32384637	32387739	-
Ncoa3	MGI:1276535	2	165992636	166073242	+
Ncoa5	MGI:2385165	2	165000357	165034867	-
Pax5	MGI:97490	2	105668900	105697364	+
Pdcd9	MGI:97419	2	143948145	143948295	+

