

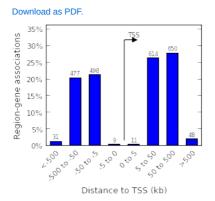
Region-Gene Association Graphs

What do these graphs illustrate?

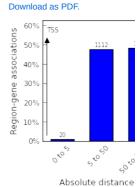
Download as PDF. Genomic regions associated with one or more genes Genomic regions not associated with any genes 120% 100% 1134 60% 60% 000 1 1 2 Number of associated genes per region

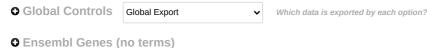
Number of associated genes per region

Binned by orientation and distance to TSS

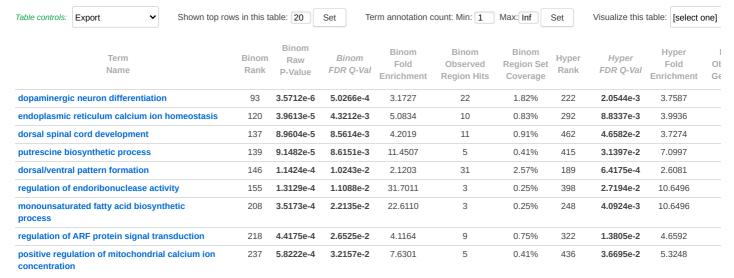


Binned by absolute distance to





- GO Biological Process (9 terms)



The test set of 1,207 genomic regions picked 2,009 (9%) of all 21,395 genes.

 $GO\ \textit{Biological Process}\ \text{has}\ 13{,}090\ \text{terms}\ \text{covering}\ 17{,}925\ (84\%)\ \text{of all}\ 21{,}395\ \text{genes},\ \text{and}\ 1{,}163{,}819\ \text{term}\ \text{-}\ \text{gene}\ \text{associations}.$

13,090 ontology terms (100%) were tested using an annotation count range of [1, Inf].

○ GO Cellular Component (1 term)

Table controls: Export
Shown top rows in this table: 20 Set Term annotation count: Min: 1 Max: Inf Set Visualize this table: [select one]

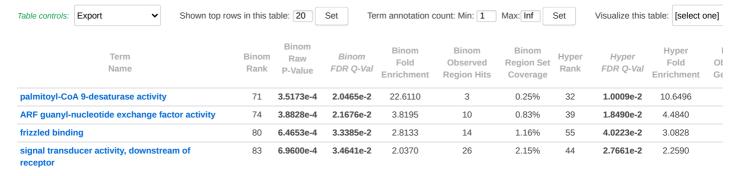


The test set of 1,207 genomic regions picked 2,009 (9%) of all 21,395 genes.

GO Cellular Component has 1,694 terms covering 19,074 (89%) of all 21,395 genes, and 371,380 term - gene associations.

1,694 ontology terms (100%) were tested using an annotation count range of [1, Inf].

○ GO Molecular Function (4 terms)



The test set of 1,207 genomic regions picked 2,009 (9%) of all 21,395 genes.

GO Molecular Function has 4,131 terms covering 17,189 (80%) of all 21,395 genes, and 227,341 term - gene associations.

4,131 ontology terms (100%) were tested using an annotation count range of [1, Inf].

- Human Phenotype (no terms)

Table controls:	Export	∨ Sh	own top rows in this to	able: 20	Set	Te	rm annotation o	count: Min: 1	Max: Inf	Set	Visualize this	table: [select one	e]
	Term Name		Binom Rank	Binom Raw P-Value	Bin FDR (Binom Fold Enrichment	Binom Observed Region Hits	Binom Region Set Coverage	Hyper Rank	Hyper FDR Q-Val		l Ol Ge
abnormal mean corpuscular volume			11	1.9207e-7	1.601	1e-4	2.4632	42	3.48%	351	3.9536e-2	1.6999	
abnormal rho	mbomere morph	nology	12	4.3977e-7	3.360	6e-4	5.5536	14	1.16%	260	1.8716e-2	4.3851	
increased ne	uron number		16	7.7990e-7	4.469	8e-4	2.9413	28	2.32%	104	4.3992e-4	3.1130	
abnormal neu	uromere morpho	logy	18	1.4471e-6	7.372	4e-4	5.0107	14	1.16%	296	2.4730e-2	4.1415	
absent supra	occipital bone		23	3.2934e-6	1.313	1e-3	6.8154	10	0.83%	331	3.3860e-2	5.3248	
decreased se	ensitivity to induc	ed cell deat	h 26	3.9699e-6	1.400	1e-3	3.1512	22	1.82%	284	2.1459e-2	2.3512	
abnormal lym	nph node cortex	morphology	35	8.3346e-6	2.183	7e-3	3.6272	17	1.41%	301	2.6028e-2	2.7782	
decreased er	ythrocyte cell nu	mber	41	1.2526e-5	2.801	.6e-3	2.1450	39	3.23%	307	2.8988e-2	1.8153	
abnormal rho	ombomere 4 mor	phology	43	1.2882e-5	2.747	1e-3	9.4133	7	0.58%	243	1.3532e-2	8.5197	
abnormal ski	n condition		44	1.3571e-5	2.828	3e-3	2.0492	43	3.56%	141	1.6561e-3	2.0708	
abnormal lymph node B cell domain morphology			46	1.6693e-5	3.327	8e-3	3.5985	16	1.33%	321	3.0884e-2	2.8572	
sparse hair			65	6.4972e-5	9.166	1e-3	2.5467	23	1.91%	287	2.1813e-2	2.2821	
abnormal opt	tic tract morphol	ogy	67	6.9342e-5	9.490	5e-3	5.3036	9	0.75%	331	3.3860e-2	5.3248	
abnormal cor	pora quadrigemi	na morphol	ogy 76	1.1987e-4	1.446	4e-2	2.8108	18	1.49%	166	3.8543e-3	3.7789	
abnormal wo	und healing		90	1.7162e-4	1.748	6e-2	2.0177	33	2.73%	365	4.4237e-2	1.8337	
increased heart weight		113	2.7456e-4	2.228	1e-2	2.0137	31	2.57%	288	2.1790e-2	1.8993		
abnormal styloid process morphology		120	3.3780e-4	2.581	4e-2	3.8891	10	0.83%	245	1.4287e-2	5.3248		
abnormal heart ventricle wall thickness		128	3.6390e-4	2.607	0e-2	2.0058	30	2.49%	202	8.5592e-3	2.1137		
abnormal ear	development		130	3.8902e-4	2.744	1e-2	2.1987	24	1.99%	125	1.2876e-3	3.2150	
increased co	chlear hair cell n	umber	147	5.5696e-4	3.474	3e-2	3.1708	12	0.99%	327	3.2589e-2	3.9235	

The test set of 1,207 genomic regions picked 2,009 (9%) of all 21,395 genes.

Mouse Phenotype Single KO has 9,170 terms covering 9,466 (44%) of all 21,395 genes, and 551,620 term - gene associations.

9,170 ontology terms (100%) were tested using an annotation count range of [1, Inf].

•	Mouse	Phenotype	(20 +	terms)
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Table controls:	Export	~	Shown top rows in this table: 20 Set	Term annotation count: Min: 1	Max: Inf Set	Visualize this table: [select one]

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Name	Rank	P-Value	FDR Q-Vai	Enrichment	Region Hits	Coverage	Rank	FDR Q-Vai	Enrichment	Ge
abnormal mean corpuscular volume	22	5.4022e-7	2.3512e-4	2.2614	46	3.81%	563	4.4649e-2	1.6083	
abnormal spinal cord dorsal horn morphology	26	1.5472e-6	5.6979e-4	6.5611	11	0.91%	236	2.7045e-3	5.0116	
increased neuron number	27	1.6767e-6	5.9461e-4	2.3755	38	3.15%	189	7.4497e-4	2.4882	
small scala media	29	2.0765e-6	6.8561e-4	6.3604	11	0.91%	380	1.6689e-2	5.9164	
absent supraoccipital bone	35	3.2934e-6	9.0098e-4	6.8154	10	0.83%	444	2.6358e-2	5.3248	
abnormal fibroblast physiology	36	4.1809e-6	1.1120e-3	2.0023	51	4.23%	340	1.0904e-2	1.7258	
increased monocyte cell number	53	1.4787e-5	2.6713e-3	2.2367	35	2.90%	428	2.2945e-2	1.8521	
abnormal rhombomere 4 morphology	60	2.2189e-5	3.5410e-3	8.6386	7	0.58%	423	2.2532e-2	7.0997	
increased sensory neuron number	63	2.4719e-5	3.7569e-3	2.5319	26	2.15%	202	1.1570e-3	3.0174	
decreased sensitivity to induced cell death	77	6.3230e-5	7.8627e-3	2.5514	23	1.91%	516	3.8043e-2	2.0836	
abnormal corpora quadrigemina morphology	111	1.5705e-4	1.3547e-2	2.6611	19	1.57%	203	1.1624e-3	3.8726	
abnormal physiological neovascularization	121	1.8127e-4	1.4345e-2	3.6010	12	0.99%	558	4.4490e-2	3.1554	
increased cardiomyocyte apoptosis	126	2.0907e-4	1.5888e-2	2.5263	20	1.66%	356	1.3125e-2	2.5765	
abnormal tectum morphology	147	2.9010e-4	1.8896e-2	2.3991	21	1.74%	191	8.3443e-4	3.5499	
abnormal cardiomyocyte apoptosis	181	5.0137e-4	2.6523e-2	2.2474	22	1.82%	396	1.8778e-2	2.3211	
increased response of heart to induced stress	188	5.5736e-4	2.8387e-2	2.5444	17	1.41%	386	1.7108e-2	2.8399	
enlarged myocardial fiber	203	7.1589e-4	3.3767e-2	2.3457	19	1.57%	211	1.4249e-3	2.8611	
sparse hair	211	7.8481e-4	3.5614e-2	2.0527	25	2.07%	507	3.5680e-2	2.0094	
abnormal styloid process morphology	215	8.2108e-4	3.6567e-2	3.2210	11	0.91%	271	4.2622e-3	5.3248	
abnormal optic tract morphology	230	9.5665e-4	3.9826e-2	3.3896	10	0.83%	482	3.2065e-2	4.2598	

The test set of 1,207 genomic regions picked 2,009 (9%) of all 21,395 genes.

Mouse Phenotype has 9,575 terms covering 9,654 (45%) of all 21,395 genes, and 705,265 term - gene associations.

9,575 ontology terms (100%) were tested using an annotation count range of [1, Inf].



Bejerano Lab



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