

Supplemental Figure S8. Interrogation of MGI

Mammalian Phenotype databases identifies genes associated with human-specific SNCs and implicated in premature death and embryonic, perinatal, neonatal, and postnatal lethality phenotypes using the Enrichr bioinformatics platform (see Methods for details).

MGI Mammalian Phenotype 2017: 8,405 genes

MP:0001262_decreased_body_weight

MP:0011087_neonatal_lethality,_complete_penetrance

MP:0001265_decreased_body_size

MP:0011086_postnatal_lethality,_incomplete_penetrance

MP:0011090_perinatal_lethality,_incomplete_penetrance

MP:0001899_absent_long_term_depression

MP:0011109_lethality_throughout_fetal_growth_and_development,_incomplete_penetrance

MP:0011085_postnatal_lethality,_complete_penetrance

MP:0002083_premature_death

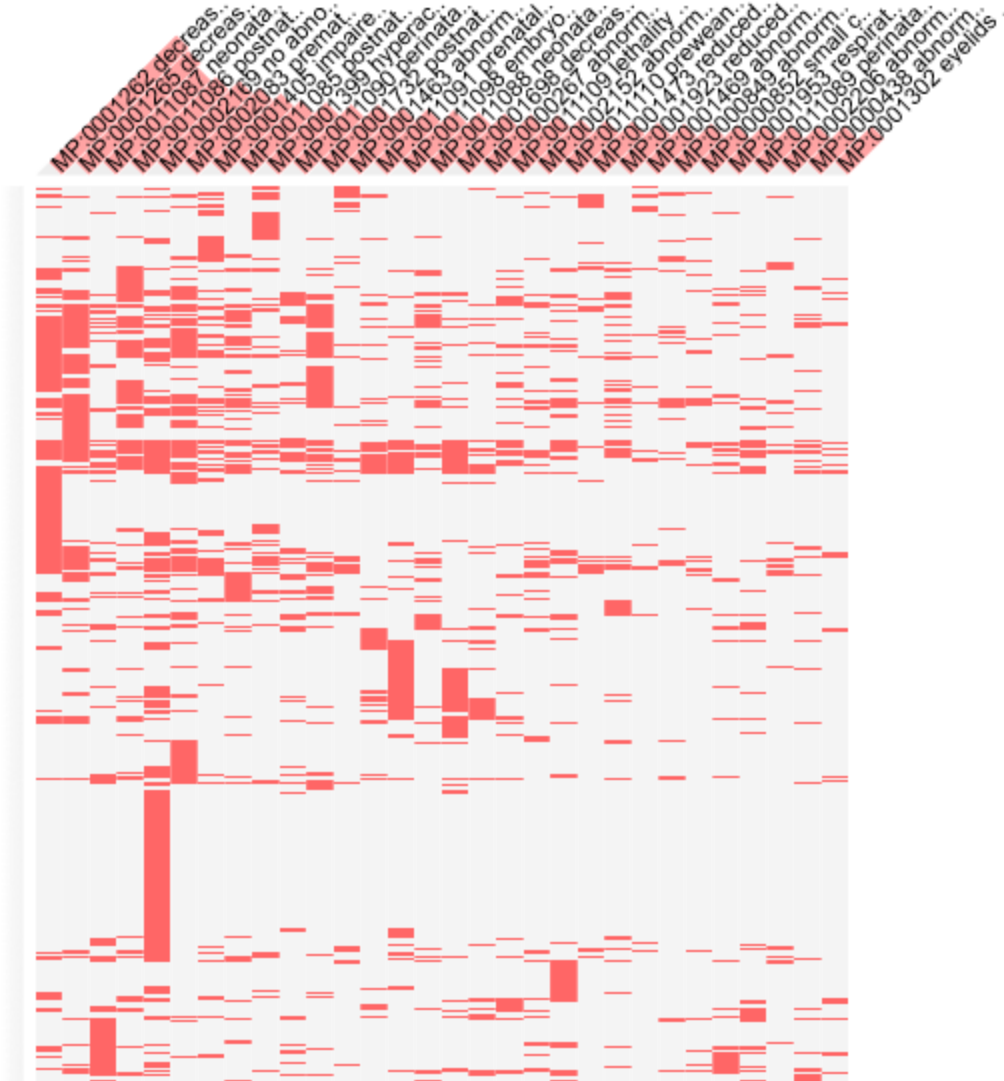
MP:0002169_no_abnormal_phenotype_detected

MGI Mammalian Phenotype 2017: 8,405 genes



Enriched Terms

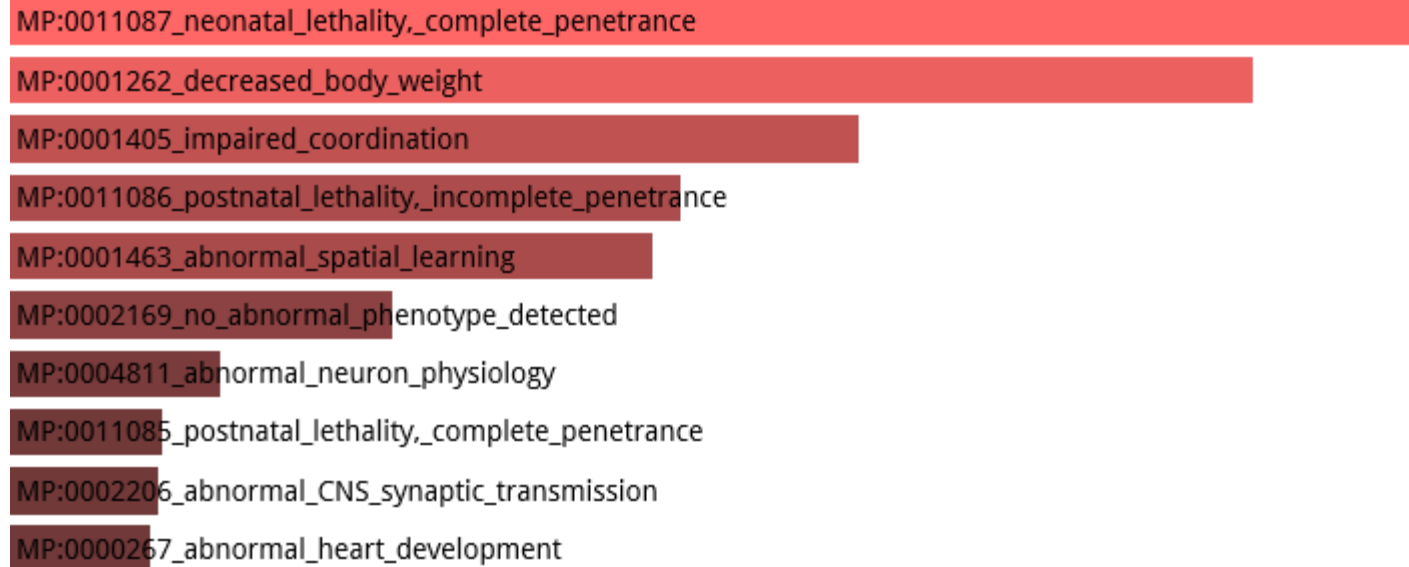
Input Genes



MGI Mammalian Phenotype 2017 (8,405 genes): Top 40 of 749 significant records

Term	Overlap	P-value	Adjusted P-value
MP:0001262_decreased_body_weight	692/1189	4.71E-31	2.44E-27
MP:0001265_decreased_body_size	472/774	2.25E-27	5.82E-24
MP:0011087_neonatal_lethality,_complete_penetrance	299/462	2.47E-23	4.25E-20
MP:0011086_postnatal_lethality,_incomplete_penetrance	346/563	4.06E-21	5.26E-18
MP:0002169_no_abnormal_phenotype_detected	882/1674	2.98E-20	3.08E-17
MP:0002083_premature_death	474/834	1.12E-18	9.65E-16
MP:0001405_impaired_coordination	215/332	3.43E-17	2.54E-14
MP:0011085_postnatal_lethality,_complete_penetrance	238/384	1.57E-15	1.02E-12
MP:0001399_hyperactivity	216/344	4.36E-15	2.51E-12
MP:0011090_perinatal_lethality,_incomplete_penetrance	152/226	1.28E-14	6.65E-12
MP:0001732_postnatal_growth_retardation	339/590	1.43E-14	6.75E-12
MP:0001463_abnormal_spatial_learning	115/162	6.87E-14	2.96E-11
MP:0011091_prenatal_lethality,_complete_penetrance	174/272	1.81E-13	7.22E-11
MP:0011098_embryonic_lethality_during_organogenesis,_complete_penetrance	319/559	2.9E-13	1.07E-10
MP:0011088_neonatal_lethality,_incomplete_penetrance	163/255	1.15E-12	3.97E-10
MP:0001698_decreased_embryo_size	273/472	1.96E-12	6.36E-10
MP:0000267_abnormal_heart_development	109/157	3.11E-12	9.48E-10
MP:0011109_lethality_throughout_fetal_growth_and_development,_incomplete_penetrance	116/170	3.92E-12	1.13E-09
MP:0002152_abnormal_brain_morphology	104/152	3.9E-11	1.06E-08
MP:0011110_preweaning_lethality,_incomplete_penetrance	222/381	8.85E-11	2.29E-08
MP:0001473_reduced_long_term_potentiation	77/106	1.52E-10	3.75E-08
MP:0001923_reduced_female_fertility	142/227	3.06E-10	7.2E-08
MP:0004811_abnormal_neuron_physiology	67/90	4.06E-10	8.85E-08
MP:0000788_abnormal_cerebral_cortex_morphology	98/145	4.1E-10	8.85E-08
MP:0001406_abnormal_gait	180/302	4.48E-10	9.28E-08
MP:0001469_abnormal_contextual_conditioning_behavior	45/54	4.96E-10	9.82E-08
MP:0000849_abnormal_cerebellum_morphology	68/92	5.12E-10	9.82E-08
MP:0000266_abnormal_heart_morphology	139/224	1.03E-09	1.91E-07
MP:0000852_small_cerebellum	52/66	1.15E-09	2.05E-07
MP:0001953_respiratory_failure	98/147	1.29E-09	2.23E-07
MP:0011089_perinatal_lethality,_complete_penetrance	135/217	1.41E-09	2.35E-07
MP:0002206_abnormal_CNS_synaptic_transmission	66/90	1.6E-09	2.59E-07
MP:0000438_abnormal_cranium_morphology	90/133	1.91E-09	2.99E-07
MP:0001302_eyelids_open_at_birth	43/52	1.97E-09	3E-07
MP:0000807_abnormal_hippocampus_morphology	63/86	4.04E-09	5.97E-07
MP:0006009_abnormal_neuronal_migration	57/76	5.14E-09	7.28E-07
MP:0001954_respiratory_distress	111/174	5.2E-09	7.28E-07
MP:0001899_absent_long_term_depression	25/26	5.88E-09	7.95E-07
MP:0002906_increased_susceptibility_to_pharmacologically_induced_seizures	65/90	5.99E-09	7.95E-07
MP:0003633_abnormal_nervous_system_physiology	74/106	6.33E-09	8.12E-07

MGI Mammalian Phenotype Level 4 2019: 8.405 genes

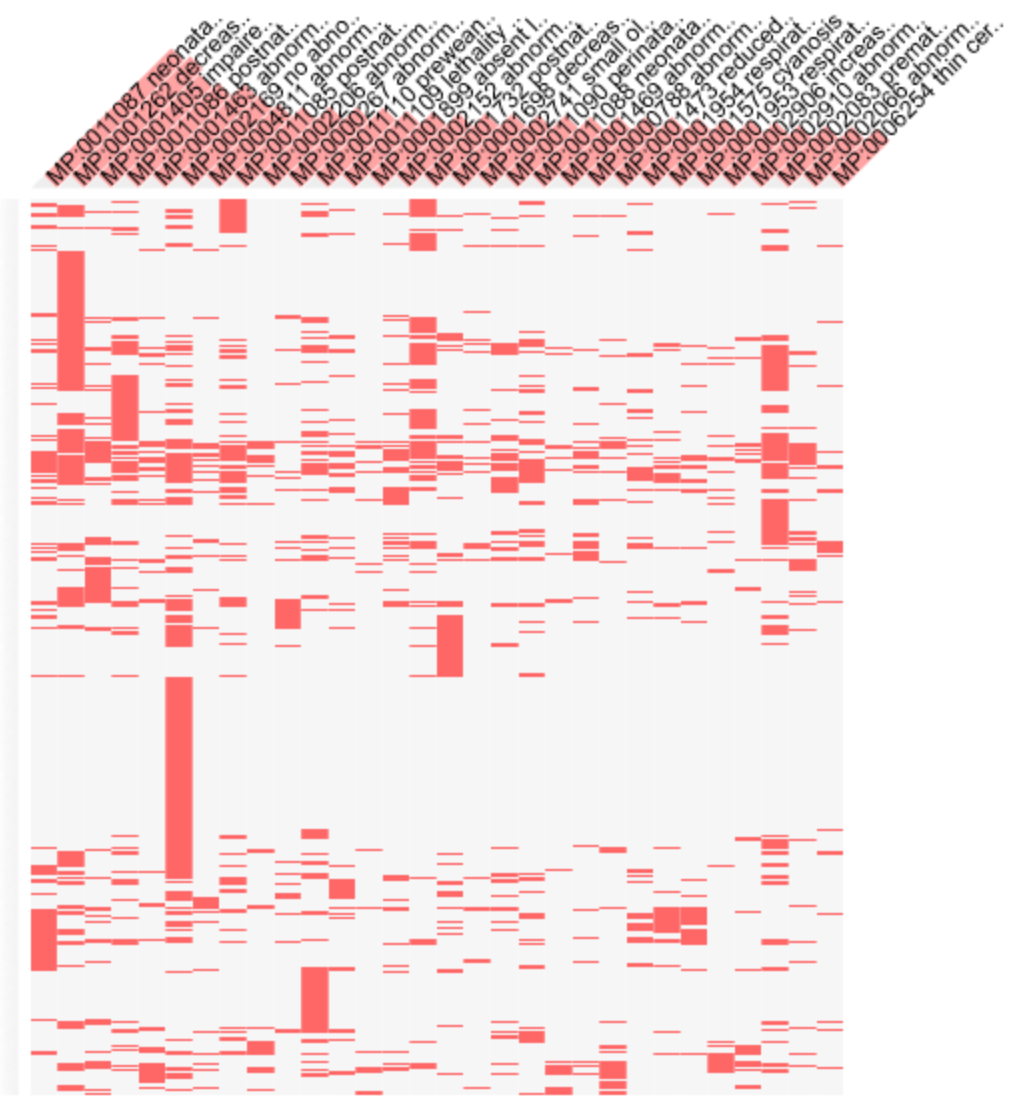


MGI Mammalian Phenotype Level 4 2019: 8.405 genes



Enriched Terms

Input Genes



MGI Mammalian Phenotype Level 4 2019 (8,405 genes): top 40 of 407 significant records

Term	Overlap	P-value	Adjusted P-value	
MP:0011087_neonatal_lethality,_complete_penetrance	315/517		1.55E-18	8.15E-15
MP:0001262_decreased_body_weight	773/1471		2.04E-17	5.36E-14
MP:0001405_impaired_coordination	247/405		6.90E-15	1.21E-11
MP:0011086_postnatal_lethality,_incomplete_penetrance	362/643		9.65E-14	1.27E-10
MP:0001463_abnormal_spatial_learning	120/172		1.46E-13	1.54E-10
MP:0002169_no_abnormal_phenotype_detected	958/1944		6.87E-12	6.02E-09
MP:0004811_abnormal_neuron_physiology	78/107		8.69E-11	6.53E-08
MP:0002206_abnormal_CNS_synaptic_transmission	75/103		2.18E-10	1.27E-07
MP:0000267_abnormal_heart_development	111/168		2.43E-10	1.28E-07
MP:0011085_postnatal_lethality,_complete_penetrance	246/432		2.04E-10	1.34E-07
MP:0011110_prewaning_lethality,_incomplete_penetrance	360/669		2.90E-10	1.39E-07
MP:0011109_lethality_throughout_fetal_growth_and_development,_incomplete_penetrance	122/191		8.21E-10	3.60E-07
MP:0001899_absent_long_term_depression	27/28		1.11E-09	4.50E-07
MP:0001732_postnatal_growth_retardation	360/677		1.85E-09	6.48E-07
MP:0001698_decreased_embryo_size	293/537		2.09E-09	6.88E-07
MP:0002152_abnormal_brain_morphology	119/187		1.84E-09	6.92E-07
MP:0011090_perinatal_lethality,_incomplete_penetrance	154/256		3.23E-09	9.44E-07
MP:0002741_small_olfactory_bulb	35/40		3.07E-09	9.51E-07
MP:0001469_abnormal_contextual_conditioning_behavior	48/61		5.45E-09	1.43E-06
MP:0001473_reduced_long_term_potentiation	84/124		6.05E-09	1.45E-06
MP:0000788_abnormal_cerebral_cortex_morphology	104/161		5.79E-09	1.45E-06
MP:0011088_neonatal_lethality,_incomplete_penetrance	172/293		5.33E-09	1.48E-06
MP:0001954_respiratory_distress	121/194		7.90E-09	1.81E-06
MP:0001575_cyanosis	129/210		1.00E-08	2.19E-06
MP:0001953_respiratory_failure	103/161		1.47E-08	3.09E-06
MP:0002906_increased_susceptibility_to_pharmacologically_induced_seizures	70/101		2.60E-08	5.25E-06
MP:0002910_abnormal_excitatory_postsynaptic_currents	59/83		7.68E-08	1.50E-05
MP:0002083_premature_death	499/997		9.80E-08	1.84E-05
MP:0002066_abnormal_motor_capabilities/coordination/movement	102/164		1.41E-07	2.55E-05
MP:0006254_thin_cerebral_cortex	53/74		2.36E-07	4.14E-05
MP:0011098_embryonic_lethality_during_organogenesis,_complete_penetrance	339/656		2.60E-07	4.28E-05
MP:0000807_abnormal_hippocampus_morphology	63/92		2.56E-07	4.34E-05
MP:0006009_abnormal_neuronal_migration	59/85		2.94E-07	4.69E-05
MP:0011108_embryonic_lethality_during_organogenesis,_incomplete_penetrance	143/247		3.20E-07	4.95E-05
MP:0000031_abnormal_cochlea_morphology	44/59		3.84E-07	5.77E-05
MP:0000852_small_cerebellum	58/84		4.91E-07	7.17E-05
MP:0002063_abnormal_learning/memory/conditioning	42/56		5.43E-07	7.52E-05
MP:0003633_abnormal_nervous_system_physiology	79/123		5.36E-07	7.62E-05
MP:0010025_decreased_total_body_fat_amount	263/498		5.94E-07	8.01E-05
MP:0009937_abnormal_neuron_differentiation	75/116		7.01E-07	9.22E-05