Geneset	stat	num.genes	pval	p.adj	gene.vals
NADH Dehydrogenase Complex Assembly (GO:	-0.34082393	49	1.587e-16	4.290e-13	TMEM126B:3 NDUFA11:49 ECSIT:63 NDUFAF1:73 NDUFB8:148 NDUFB5:190
Mitochondrial Respiratory Chain Complex	-0.34082393	49	1.587e-16	4.290e-13	TMEM126B:3 NDUFA11:49 ECSIT:63 NDUFAF1:73 NDUFB8:148 NDUFB5:190
Mitochondrial Gene Expression (GO:014005	-0.23505441	102	2.542e-16	4.582e-13	TFAM:14 TEFM:93 POLRMT:101 TFB2M:112 TWNK:217 MRPS14:376
Mitochondrial Translation (GO:0032543)	-0.22711501	97	1.150e-14	1.555e-11	MRPS14:376 GADD45GIP1:477 FASTKD2:485 MRPS9:488 MRPS5:509 MRPL22:559
Mitochondrial ATP Synthesis Coupled Elec	-0.27220331	65	3.343e-14	3.615e-11	NDUFAF1:73 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422 NDUFB3:451
Aerobic Electron Transport Chain (GO:001	-0.27107537	64	6.639e-14	5.983e-11	NDUFAF1:73 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422 NDUFB3:451
Mitochondrial Respiratory Chain Complex	-0.23698959	82	1.244e-13	9.608e-11	TMEM126B:3 NDUFA11:49 ECSIT:63 NDUFAF1:73 NDUFB8:148 NDUFB5:190
Intermediate Filament Organization (GO:0	0.25637844	65	9.107e-13	6.155e-10	PKP1:8 DSP:30 KRT17:105 KRT80:119 KRT20:129 DES:220
Monoatomic Cation Transmembrane Transpor	0.12346999	277	1.821e-12	1.094e-09	TRPM5:13 TRPV6:31 SLC24A3:125 KCNQ3:126 CACNA1S:152 SLC24A4:169
Oxidative Phosphorylation (GO:0006119)	-0.26105936	59	4.166e-12	2.252e-09	NDUFA11:49 TEFM:93 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422
Proton Motive Force-Driven Mitochondrial	-0.27879639	50	9.319e-12	4.581e-09	NDUFA11:49 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422 NDUFB3:451
Translation (GO:0006412)	-0.13473174	208	2.376e-11	1.071e-08	MRPS14:376 ZAR1:424 HBS1L:440 GADD45GIP1:477 FASTKD2:485 MRPS9:488
Mitochondrial Electron Transport, NADH T	-0.32843823	33	6.670e-11	2.774e-08	NDUFAF1:73 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422 NDUFB3:451
Proton Motive Force–Driven ATP Synthesis	-0.25205853	55	1.030e-10	3.976e-08	NDUFA11:49 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422 NDUFB3:451
Chemical Synaptic Transmission (GO:00072	0.11476942	263	1.697e-10	6.118e-08	KCNQ3:126 HRH2:130 CACNA1E:166 GRIK4:236 HTR5A:246 SLC18A3:251
Inorganic Cation Transmembrane Transport	0.11013440	280	2.622e-10	8.860e-08	TRPM5:13 TRPV6:31 SLC24A3:125 KCNQ3:126 CACNA1S:152 SLC24A4:169
Cilium Organization (GO:0044782)	-0.12170787	221	5.042e-10	1.604e-07	KIAA0586:44 KIF27:46 ARL13B:87 GBF1:126 CENPJ:149 IQCG:231
tRNA Modification (GO:0006400)	-0.21526005	67	1.137e-09	3.416e-07	MTO1:174 TYW5:244 PUS10:301 TRMT10A:361 METTL8:364 DTWD1:394
Cilium Assembly (GO:0060271)	-0.11396835	232	2.474e-09	7.040e-07	TBC1D32:31 KIAA0586:44 KIF27:46 ARL13B:87 GBF1:126 CENPJ:149
Muscle Contraction (GO:0006936)	0.17646929	91	6.161e-09	1.666e-06	MYH2:11 MYH13:54 MYOF:127 CACNA1S:152 DES:220 MYH1:239
Cellular Respiration (GO:0045333)	-0.18370358	82	9.167e-09	2.360e-06	NDUFA11:49 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422 NDUFB3:451
Aerobic Respiration (GO:0009060)	-0.21956862	57	1.001e-08	2.460e-06	NDUFA11:49 TEFM:93 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422
Calcium Ion Transmembrane Import Into Cy	0.18574261	79	1.180e-08	2.774e-06	TRPV6:31 CACNA1I:148 CACNA1S:152 CACNA1E:166 SLC24A4:169 CACNA1G:287
Calcium Ion Transport (GO:0006816)	0.15520251	110	1.950e-08	4.393e-06	TRPM5:13 TRPV6:31 SLC24A3:125 CACNA1S:152 SLC24A4:169 CACNA1G:287
Sensory Perception Of Smell (GO:0007608)	0.11040761	208	4.383e-08	9.481e-06	SLC24A4:169 OR10A2:211 OR2H2:369 OR10D3:475 DRD2:522 OR10C1:636
Potassium Ion Transmembrane Transport (G	0.13547872	135	5.751e-08	1.196e-05	SLC24A3:125 KCNQ3:126 SLC24A4:169 KCNK10:225 KCNJ12:259 KCNH2:365
Potassium Ion Transport (GO:0006813)	0.13631751	121	2.319e-07	4.645e-05	SLC24A3:125 KCNQ3:126 SLC24A4:169 KCNK10:225 KCNJ12:259 KCNH2:365
Positive Regulation Of DNA Metabolic Pro	-0.14608275	104	2.729e-07	5.269e-05	RNF168:52 STON1:61 SMCHD1:136 ABRAXAS1:141 DNA2:193 UIMC1:291
Response To Calcium Ion (GO:0051592)	0.14622412	100	4.487e-07	8.365e-05	SYT2:18 TRPV6:31 ADCY1:32 SYT8:179 RASGRP2:240 SYT6:275
Neuron Projection Morphogenesis (GO:0048	0.12392007	139	4.761e-07	8.582e-05	ADGRB1:39 NRCAM:96 CTTN:149 TRIO:200 FLRT1:218 TNN:252
DNA Repair (GO:0006281)	-0.08740794	281	5.052e-07	8.811e-05	RNF168:52 STON1:61 USP44:64 TOPBP1:123 ABRAXAS1:141 DNA2:193
Cardiac Conduction (GO:0061337)	0.21419307	45	6.722e-07	1.136e-04	DSP:30 DSG2:104 CACNA1G:287 KCNH2:365 DSC2:569 CACNB2:890
Regulation Of Heart Rate By Cardiac Cond	0.22627703	40	7.413e-07	1.215e-04	DSP:30 DSG2:104 CACNA1G:287 KCNH2:365 DSC2:569 CACNB2:890
tRNA Methylation (GO:0030488)	-0.23693916	36	8.735e-07	1.389e-04	MTO1:174 TRMT61B:285 TRMT10C:293 TRMT10A:361 METTL8:364 FTSJ1:505
Anterograde Trans-Synaptic Signaling (GO	0.10311036	190	1.010e-06	1.560e-04	KCNQ3:126 HRH2:130 CACNA1E:166 GRIK4:236 HTR5A:246 SLC18A3:251
Modulation Of Chemical Synaptic Transmis	0.12903473	118	1.332e-06	2.001e-04	MYOF:127 TRIO:200 GRIK4:236 OTOF:272 GRM2:438 DRD2:522
Calcium Ion Transmembrane Transport (GO:	0.15800063	78	1.434e-06	2.095e-04	TRPM5:13 TRPV6:31 SLC24A3:125 CACNA1S:152 SLC24A4:169 CACNA1G:287
Cellular Response To Calcium Ion (GO:007	0.16811948	67	1.980e-06	2.818e-04	SYT2:18 ADCY1:32 SYT8:179 RASGRP2:240 SYT6:275 CAPN3:448
Metal Ion Transport (GO:0030001)	0.10488688	169	2.671e-06	3.702e-04	SLC17A4:9 TRPV6:31 CACNA1S:152 KCNJ12:259 CHRNA4:393 ITPR1:416
Mitochondrial RNA Metabolic Process (GO:	-0.29839148	20	3.852e-06	5 207e-04	TFAM:14 TEFM:93 POLRMT:101 TFB2M:112 TWNK:217 FASTKD1:431

EnrichmentHsSymbolsFile2 Top pathways by non-permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
REACTOME_KERATINIZATION	0.22485940	214	9.785e-30	6.355e-26	PKP1:8 DSP:30 KRTAP6-1:34 KRTAP10-1:83 KRTAP10-3:84 DSG2:104
FISCHER_DREAM_TARGETS	-0.10069796	924	5.494e-25	1.784e-21	DLGAP5:10 ATAD2:27 PARPBP:29 KIF18A:30 CEP192:39 SGO2:62
JOHNSTONE_PARVB_TARGETS_3_DN	-0.09707511	794	2.019e-20	4.369e-17	CCPG1:1 UBAP2:5 FAM13B:6 SRFBP1:8 DLGAP5:10 TFAM:14
MARSON_BOUND_BY_E2F4_UNSTIMULATED	-0.10201173	664	4.108e-19	6.670e-16	UBAP2:5 DLGAP5:10 PARPBP:29 LRRC40:33 SGO2:62 DNMT1:74
REACTOME_NEURONAL_SYSTEM	0.13056085	394	6.861e-19	8.911e-16	SHANK2:3 SYT2:18 PPFIA1:23 ADCY1:32 GJA10:91 PPFIA4:107
REACTOME_FORMATION_OF_THE_CORNIFIED_ENVE	0.22527613	128	1.407e-18	1.522e-15	PKP1:8 DSP:30 DSG2:104 KRT17:105 KRT80:119 KRT20:129
REACTOME_SENSORY_PERCEPTION	0.10279761	603	8.354e-18	7.750e-15	TRPM5:13 RGS9:52 MYO15A:68 OR10A2:211 OR51E2:230 OTOF:272
HAMAI_APOPTOSIS_VIA_TRAIL_UP	-0.09519660	625	5.595e-16	4.542e-13	FAM13B:6 DLGAP5:10 CEP350:22 ATAD2:27 PARPBP:29 KIF18A:30
RODRIGUES_THYROID_CARCINOMA_POORLY_DIFFE	-0.09655581	601	7.640e-16	5.513e-13	CCPG1:1 DLGAP5:10 LRPPRC:13 TFAM:14 ATAD2:27 PARPBP:29
MARTENS_TRETINOIN_RESPONSE_UP	0.08438836	788	1.041e-15	5.647e-13	SHANK2:3 CEP170B:19 TRIM29:35 AXIN2:98 JAG2:138 RNF186:139
REACTOME_RESPIRATORY_ELECTRON_TRANSPORT	-0.24471771	90	1.043e-15	5.647e-13	TMEM126B:3 LRPPRC:13 NDUFA11:49 TIMMDC1:56 ECSIT:63 NDUFAF1:73
DODD_NASOPHARYNGEAL_CARCINOMA_DN	-0.06723991	1278	1.042e-15	5.647e-13	TET1:2 DLGAP5:10 LRPPRC:13 TFAM:14 MTPAP:25 ATAD2:27
WP_MITOCHONDRIAL_COMPLEX_I_ASSEMBLY_MODE	-0.33691063	47	1.345e-15	6.720e-13	TMEM126B:3 TIMMDC1:56 ECSIT:63 NDUFAF1:73 TMEM70:95 NDUFB8:148
REACTOME_DEVELOPMENTAL_BIOLOGY	0.07004333	1097	7.012e-15	3.253e-12	PKP1:8 DSP:30 KRTAP6-1:34 SEMA7A:58 KRTAP10-1:83 KRTAP10-3:84
REACTOME_MITOCHONDRIAL_TRANSLATION	-0.23115179	93	1.344e-14	5.820e-12	MRPS35:322 MRPS14:376 GADD45GIP1:477 MRPS9:488 MRPS5:509 MRPL22:559
REACTOME_RESPIRATORY_ELECTRON_TRANSPORT_	-0.20979641	112	1.764e-14	6.737e-12	TMEM126B:3 LRPPRC:13 NDUFA11:49 TIMMDC1:56 ECSIT:63 NDUFAF1:73
REACTOME_COMPLEX_I_BIOGENESIS	-0.31996423	48	1.739e-14	6.737e-12	TMEM126B:3 NDUFA11:49 TIMMDC1:56 ECSIT:63 NDUFAF1:73 NDUFB8:148
REACTOME_SIGNALING_BY_GPCR	0.08400692	687	7.518e-14	2.712e-11	GPR37L1:12 TAAR6:28 ADCY1:32 RGS9:52 CALCA:63 TIAM1:111
WP_GPCRS_CLASS_A_RHODOPSINLIKE	0.13432814	253	2.006e-13	6.856e-11	GPR37L1:12 HRH2:130 FFAR2:150 GPR68:174 GPER1:188 GPR35:205
MEISSNER_NPC_HCP_WITH_H3K4ME2_AND_H3K27M	0.11476644	340	3.870e-13	1.256e-10	SYT2:18 BNC1:26 ADGRB1:39 LAD1:42 SEMA7A:58 JAG2:138
MIKKELSEN_MEF_HCP_WITH_H3K27ME3	0.08850454	577	4.363e-13	1.349e-10	ADGRB1:39 LAD1:42 DSG2:104 KCNQ3:126 HRH2:130 JAG2:138
REACTOME_CELL_CYCLE	-0.08305517	652	5.582e-13	1.648e-10	DAXX:12 KIF18A:30 CEP192:39 SYNE2:43 RNF168:52 SGO2:62
KEGG_CALCIUM_SIGNALING_PATHWAY	0.15946070	168	1.036e-12	2.925e-10	ADCY1:32 HRH2:130 CACNA1I:148 CACNA1S:152 CACNA1E:166 HTR5A:246
MIKKELSEN_MCV6_HCP_WITH_H3K27ME3	0.10049285	426	1.263e-12	3.419e-10	SYT2:18 ADGRB1:39 SEMA7A:58 ZBTB16:76 KCNQ3:126 CLMP:143
BENPORATH_ES_WITH_H3K27ME3	0.06463374	1063	1.462e-12	3.797e-10	PKP1:8 PLEC:15 BNC1:26 PHLDB1:45 RGS9:52 CALCA:63
KINSEY_TARGETS_OF_EWSR1_FLII_FUSION_UP	-0.06071051	1204	1.845e-12	4.608e-10	DLGAP5:10 ATAD2:27 PARPBP:29 KIF18A:30 LARP4:38 SGO2:62
NIKOLSKY_BREAST_CANCER_7P22_AMPLICON	0.33247509	37	2.593e-12	6.233e-10	INTS1:109 GPER1:188 ZFAND2A:199 SNX8:387 MAD1L1:459 AP5Z1:508
REACTOME_MUSCLE_CONTRACTION	0.14500232	196	2.687e-12	6.233e-10	MYBPC2:144 MME:145 CASQ1:147 CACNA1I:148 DES:220 KCNK10:225
REACTOME_OLFACTORY_SIGNALING_PATHWAY	0.10246751	390	4.139e-12	9.269e-10	OR10A2:211 OR51E2:230 OR4K15:315 OR2H2:369 OR9K2:400 OR10D3:475
GOBERT_OLIGODENDROCYTE_DIFFERENTIATION_U	-0.08599122	558	4.358e-12	9.433e-10	DLGAP5:10 ATAD2:27 PARPBP:29 KIF18A:30 CEP192:39 KIAA0586:44
YOSHIMURA_MAPK8_TARGETS_UP	0.05905172	1226	4.813e-12	1.008e-09	AFDN:4 SYT2:18 ADH7:37 ARC:49 RGS9:52 CALCA:63
KEGG_OLFACTORY_TRANSDUCTION	0.10253300	377	8.933e-12	1.813e-09	OR10A2:211 OR51E2:230 OR4K15:315 OR2H2:369 OR9K2:400 OR6B3:501
FLORIO_NEOCORTEX_BASAL_RADIAL_GLIA_DN	-0.14435381	186	1.170e-11	2.302e-09	DLGAP5:10 PARPBP:29 KIF18A:30 SGO2:62 KIF22:90 KIF2C:108
WP_2Q37_COPY_NUMBER_VARIATION_SYNDROME	0.17186962	129	1.613e-11	3.080e-09	PER2:2 CALCA:63 PASK:65 KIF1A:161 GPR35:205 PPP1R7:244
BLANCO_MELO_BRONCHIAL_EPITHELIAL_CELLS_I	-0.14567517	175	3.083e-11	5.720e-09	KIF2C:108 SPAG5:163 NEXN:164 GCNT3:201 FANCG:202 KIF20A:251
MIKKELSEN_NPC_HCP_WITH_H3K27ME3	0.10550387	335	3.454e-11	6.230e-09	SYT2:18 BNC1:26 ADGRB1:39 LAD1:42 JAG2:138 LMX1A:306
REACTOME_CELL_CYCLE_CHECKPOINTS	-0.11719376	270	3.598e-11	6.315e-09	KIF18A:30 RNF168:52 SGO2:62 CENPC:70 CENPT:71 KIF2C:108
KEGG_NEUROACTIVE_LIGAND_RECEPTOR_INTERAC	0.11650318	265	7.010e-11	1.198e-08	TAAR6:28 HRH2:130 GPR35:205 GRIK4:236 HTR5A:246 ADORA2A:269
	0.400====:				NEUTRO 440 NEUTRO 400 NEUTRO 440

DisGeNET Top pathways by non-permulation

3.619e-10 5.875e-08

438

9.209e-11 1.533e-08 NDUFB8:148 NDUFB5:190 NDUFB10:418 NDUFS5:422 NDUFB3:451 NDUFB4:502

DLGAP5:10 LRPPRC:13 MTPAP:25 ATAD2:27 PARPBP:29 KIF18A:30

WP_ELECTRON_TRANSPORT_CHAIN_OXPHOS_SYSTE -0.19876994

SHEDDEN_LUNG_CANCER_POOR_SURVIVAL_A6 -0.08756792

Geneset	stat	num.genes	pval	p.adj	gene.vals
Schizophrenia	0.05648410	1731	2.403e-14	2.359e-10	SHANK2:3 SORL1:5 TRANK1:7 PLEC:15 NAV1:17 TAAR6:28
Arthrogryposis	0.09909345	298	4.549e-09	2.233e-05	MYH2:11 PLEC:15 SYT2:18 OFD1:121 COL6A1:158 IGHMBP2:176
Bipolar Disorder	0.06247038	755	7.235e-09	2.368e-05	PER2:2 SORL1:5 TRANK1:7 TAAR6:28 TRPV6:31 ADCY1:32
Increased CSF lactate	-0.21571940	56	2.394e-08	5.876e-05	TMEM126B:3 LRPPRC:13 NDUFA11:49 TIMMDC1:56 NDUFAF1:73 TRMT10C:293
Charcot-Marie-Tooth Disease	0.09819164	268	3.482e-08	6.837e-05	SYT2:18 DSP:30 SBF1:33 PRKN:57 MYO15A:68 DSG2:104
NADH:Q(1) Oxidoreductase deficiency	-0.30491833	25	1.317e-07	1.847e-04	TMEM126B:3 NDUFA11:49 TIMMDC1:56 NDUFAF1:73 NDUFB10:418 NDUFB3:451
Primary microcephaly	-0.14646853	110	1.154e-07	1.847e-04	CENPJ:149 DNA2:193 FANCG:202 STIL:219 XRCC4:255 KNL1:350
Autism Spectrum Disorders	0.06649237	516	2.808e-07	2.757e-04	SHANK2:3 MKI67:10 PRKN:57 KMT5B:61 PPP1R13B:64 ZNF423:80
Autistic Disorder	0.06024552	637	2.642e-07	2.757e-04	SHANK2:3 SDK1:20 HTT:46 PRKN:57 ZNF407:60 CALCA:63
Mitochondrial Diseases	-0.07909509	363	2.566e-07	2.757e-04	TMEM126B:3 LRPPRC:13 MTPAP:25 NDUFA11:49 ECSIT:63 NDUFAF1:73
Reflex, Deep Tendon, Absent	0.12974571	129	3.748e-07	3.067e-04	SYT2:18 SBF1:33 MME:145 KIF1A:161 IGHMBP2:176 BICD2:295
Absent reflex	0.12974571	129	3.748e-07	3.067e-04	SYT2:18 SBF1:33 MME:145 KIF1A:161 IGHMBP2:176 BICD2:295
Congenital myopathy (disorder)	0.14018054	109	4.416e-07	3.335e-04	MYH2:11 CASQ1:147 CACNA1S:152 COL6A1:158 IGHMBP2:176 DES:220
Absent tendon reflex	0.12988190	126	4.964e-07	3.482e-04	SYT2:18 SBF1:33 MME:145 KIF1A:161 IGHMBP2:176 BICD2:295
Mood Disorders	0.07987856	319	1.029e-06	6.735e-04	ZHX3:24 TAAR6:28 ADCY1:32 HTT:46 PPP1R13B:64 ADARB1:157
MITOCHONDRIAL COMPLEX I DEFICIENCY	-0.27270922	26	1.491e-06	9.151e-04	TMEM126B:3 NDUFA11:49 TIMMDC1:56 NDUFAF1:73 NDUFB3:451 FOXRED1:619
Congenital pes cavus	0.12653047	120	1.745e-06	9.517e-04	SYT2:18 ANOS1:77 MME:145 HSD17B4:320 L1CAM:326 INF2:410
Unipolar Depression	0.06365788	487	1.737e-06	9.517e-04	ZHX3:24 HTT:46 ZBTB16:76 ACSL4:92 ADARB1:157 TRIO:200
Ciliopathies	-0.10474399	174	1.969e-06	1.018e-03	TBC1D32:31 KIAA0586:44 ARL13B:87 NME8:273 NPHP3:306 BUB1B:318
Gait, Drop Foot	0.25007073	30	2.142e-06	1.052e-03	IGHMBP2:176 L1CAM:326 INF2:410 MPZ:602 GNE:649 TTN:825
Abnormal mitochondria in muscle tissue	-0.27011667	25	2.950e-06	1.379e-03	TMEM126B:3 NDUFA11:49 TIMMDC1:56 NDUFAF1:73 NDUFB10:418 NDUFB3:451
Acute necrotizing encephalopathy	-0.30034987	20	3.327e-06	1.485e-03	TMEM126B:3 NDUFA11:49 TIMMDC1:56 NDUFAF1:73 NDUFB3:451 FOXRED1:619
Myopathy	0.07025291	358	5.469e-06	2.335e-03	MYH2:11 PLEC:15 CASQ1:147 COL6A1:158 IGHMBP2:176 DES:220
Abnormal behavior	0.07148187	341	6.270e-06	2.565e-03	SHANK2:3 HTT:46 NRCAM:96 ADARB1:157 HTR5A:246 HDAC4:291
Myocardial Infarction	0.04480251	896	6.918e-06	2.717e-03	SBF1:33 ARC:49 ALMS1:103 PC:185 GPER1:188 NDST1:204
Congestive heart failure	0.04714750	791	8.074e-06	2.936e-03	DSP:30 TRPV6:31 HTT:46 ARC:49 AIMP2:74 ALMS1:103
Spasmodic torticollis	0.26362179	24	7.827e-06	2.936e-03	COL6A1:158 KCTD17:368 DRD2:522 PNKD:561 CACNA1B:716 SCP2:1254
Mental Depression	0.05666113	534	8.616e-06	3.022e-03	PER2:2 ADCY1:32 HTT:46 ACSL4:92 KRT20:129 GRIK4:236
Foot-drop	0.20237383	40	9.539e-06	3.122e-03	MME:145 IGHMBP2:176 DES:220 INF2:410 MPZ:602 GNE:649
Heart failure	0.04858939	732	9.225e-06	3.122e-03	DSP:30 HTT:46 ARC:49 AIMP2:74 ALMS1:103 MME:145
Chromosome Breakage	-0.20190579	40	1.000e-05	3.168e-03	FANCG:202 XRCC4:255 BRCA1:300 FANCM:414 DNMT3B:780 IL4:927
Alzheimer's Disease	0.03187802	1795	1.231e-05	3.777e-03	PER2:2 SHANK2:3 SORL1:5 HTT:46 ARC:49 LGR6:51
nervous system disorder	0.06332214	407	1.294e-05	3.850e-03	SHANK2:3 HTT:46 CALCA:63 ACKR3:254 NOTCH1:258 ATP7B:273
Distal muscle weakness	0.14617587	74	1.397e-05	4.023e-03	SYT2:18 COL6A1:158 IGHMBP2:176 DES:220 INF2:410 MPZ:602
Torticollis	0.24585795	26	1.434e-05	4.023e-03	COL6A1:158 KCTD17:368 DRD2:522 PNKD:561 CACNA1B:716 DNM2:1182
Depressive disorder	0.04914697	678	1.522e-05	4.150e-03	PER2:2 TAAR6:28 ADCY1:32 HTT:46 AIMP2:74 ANOS1:77
Distal sensory impairment	0.15060127	68	1.774e-05	4.709e-03	SBF1:33 MME:145 KIF1A:161 IGHMBP2:176 PIK3R5:363 INF2:410
Major Depressive Disorder	0.05651225	486	2.226e-05	5.670e-03	TAAR6:28 HTT:46 ZBTB16:76 ADARB1:157 GRIK4:236 ACKR3:254
Pervasive Development Disorder	0.11216959	120	2.252e-05	5.670e-03	SHANK2:3 HTT:46 PRKN:57 KMT5B:61 TRIO:200 KMT2C:227
Distal limb muscle weakness due to perip	0.14125547	75	2.376e-05	5.832e-03	SYT2:18 COL6A1:158 IGHMBP2:176 DES:220 INF2:410 MPZ:602

MGI_Mammalian_Phenotype_Level_4 Top pathways by non-permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
MP0003635 abnormal synaptic transmissio	0.09019531	425		2.874e-07	SYT2:18 ADCY1:32 ARC:49 NRCAM:96 ADARB1:157 CACNA1E:166
MP0001929 abnormal gametogenesis	-0.08306070	470	2.406e-09	5.727e-07	ARID4A:86 FGD2:145 ADCY10:173 FANCG:202 SYCP1:216 EIF4G3:234
MP0003698 abnormal male reproductive	-0.06637641	521	5.859e-07	9.296e-05	CATSPER2:41 CLGN:84 ADCY10:173 FANCG:202 SYCP1:216 EIF4G3:234
MP0002066 abnormal motor capabilities/c	0.04659325	1087	1.822e-06	1.497e-04	PER2:2 SYT2:18 HTT:46 NRCAM:96 HRH2:130 CACNA1S:152
MP0002063 abnormal learning/memory/cond	0.07591129	356	1.590e-06	1.497e-04	PER2:2 ADCY1:32 HTT:46 ARC:49 NRCAM:96 HTR5A:246
MP0009745 abnormal behavioral response	0.09790641	205	1.887e-06	1.497e-04	PER2:2 CALCA:63 SLC18A3:251 ADORA2A:269 CACNA1G:287 UPP1:377
MP0010094 abnormal chromosome stability	-0.16744916	65	3.340e-06	2.271e-04	FANCG:202 XRCC4:255 BRCA1:300 MBD1:313 BUB1B:318 CENPE:392
MP0002085 abnormal embryonic tissue	-0.05180204	655	1.610e-05	9.580e-04	ADAMTS20:9 CUBN:11 DAXX:12 TFAM:14 NSD1:24 ECSIT:63
MP0002067 abnormal sensory capabilities	0.07410385	267	4.273e-05	2.034e-03	HTT:46 CACNA1S:152 CACNA1E:166 IGHMBP2:176 GRIK4:236 HDAC4:29
MP0002064 seizures	0.07614848	255	3.886e-05	2.034e-03	HTT:46 KCNQ3:126 HRH2:130 ADARB1:157 GRIK4:236 SLC18A3:251
MP0003077 abnormal cell cycle	-0.09570616	153	5.262e-05	2.129e-03	ITGB1:152 RIF1:179 MED1:208 BRCA1:300 BUB1B:318 CENPE:392
MP0002572 abnormal emotion/affect behav	0.07068146	287	5.366e-05	2.129e-03	ADCY1:32 HTT:46 ARC:49 ULK2:155 CACNA1E:166 HTR5A:246
MP0002210 abnormal sex determination	-0.06338802	350	7.003e-05	2.564e-03	FANCG:202 SYCP1:216 EIF4G3:234 BRCA1:300 MAGEL2:373 RNF17:386
MP0005620 abnormal muscle contractility	0.08102015	206	7.733e-05	2.579e-03	CASQ1:147 CACNA1S:152 COL6A1:158 DES:220 TNNT2:380 APOE:461
MP0000653 abnormal sex gland	-0.05226620	523	8.129e-05	2.579e-03	AFP:53 ARID4A:86 FANCG:202 SYCP1:216 EIF4G3:234 BRCA1:300
MP0002429 abnormal blood cell	-0.03635617	1240	9.287e-05	2.763e-03	SPIC:40 ARID4A:86 NR2C1:99 IL23R:113 PTPN12:131 ITGB1:152
MP0002272 abnormal nervous system	0.08022435	197	1.279e-04	3.581e-03	PLEC:15 ARC:49 NRCAM:96 KCNQ3:126 GRIK4:236 CACNA1G:287
MP0002086 abnormal extraembryonic tissu	-0.05156409	486	1.690e-04	4.469e-03	DLGAP5:10 CUBN:11 DAXX:12 NSD1:24 NANOG:65 BPTF:66
MP0000689 abnormal spleen morphology	-0.04830224	552	1.911e-04	4.548e-03	SPIC:40 DNMT1:74 ARID4A:86 GRB14:153 IKZF3:256 SERPINI2:302
MP0000313 abnormal cell death	-0.05242775	463	1.831e-04	4.548e-03	DAXX:12 TFAM:14 DNMT1:74 CTH:116 UBR5:127 ITGB1:152
MP0000749 muscle degeneration	0.17594437	36	2.677e-04	6.067e-03	PLEC:15 MYOF:127 CACNA1S:152 IGHMBP2:176 DES:220 PFKM:388
MP0000598 abnormal liver morphology	-0.04992233	482		6.180e-03	XIRP2:26 ARID4A:86 CTH:116 PTPN12:131 ITGB1:152 GRB14:153
MP0001119 abnormal female reproductive	-0.06010604	295		1.039e-02	AFP:53 ARID4A:86 SMCHD1:136 FANCG:202 SYCP1:216 BRCA1:300
MP0000716 abnormal immune system	-0.03467999	1013	5.457e-04	1.082e-02	SPIC:40 ARID4A:86 IL23R:113 PTPN12:131 NFATC3:156 CRLF2:158
MP0001145 abnormal male reproductive	-0.04806045	457	6.514e-04	1.240e-02	FANCG:202 SYCP1:216 EIF4G3:234 BRCA1:300 AGTPBP1:324 RNF17:386
MP0001970 abnormal pain threshold	0.07835581	152		1.770e-02	ADCY1:32 CALCA:63 HOXB13:88 KIF1A:161 ADORA2A:269 HDAC4:291
MP0002723 abnormal immune serum	-0.03886331	649	1.269e-03		IL23R:113 PTPN12:131 CRLF2:158 IL27RA:197 GCNT3:201 IKZF3:256
MP0002135 abnormal kidney morphology	-0.04537609	455	1.316e-03		TET2:4 CUBN:11 ARID4A:86 IKZF3:256 BRCA1:300 NPHP3:306
MP0001800 abnormal humoral immune	-0.05077512	333		3.053e-02	CRLF2:158 GCNT3:201 IKZF3:256 GABPB2:346 SH2D3C:405 REV3L:416
MP0004957 abnormal blastocyst morpholog	-0.09936407	82	1.978e-03		NANOG:65 ITGB1:152 CHD8:367 AHCTF1:402 NUSAP1:429 LAMA1:434
MP0002084 abnormal developmental patter	-0.04782237	366		3.366e-02	CUBN:11 DAXX:12 NSD1:24 ECSIT:63 NANOG:65 BPTF:66
MP0001486 abnormal startle reflex	0.08896226	96		4.042e-02	LOXHD1:128 OTOF:272 APOE:461 NTSR1:518 DRD2:522 CACNA1B:716
MP0002876 abnormal thyroid physiology	-0.19828553	19		4.042e-02	MED1:208 GHR:499 CGA:1054 AIRE:1447 GLIS3:1571 GFRA4:2849
MP0002080 prenatal lethality	-0.02699920	1335		4.057e-02	CUBN:11 DAXX:12 TFAM:14 NSD1:24 SPIC:40 ECSIT:63
MP0004085 abnormal heartbeat	0.06806791	160		4.353e-02	
MP0003693 abnormal embryo hatching	-0.17361362	24		4.353e-02	VEZT:103 KIF11:475 RBBP8:507 CDC25A:1235 CDCA8:1236 PARG:2207
MP0000579 abnormal nail morphology	0.18433457	21		4.497e-02	KRT17:105 ITGB4:223 L1CAM:326 EGFR:686 KRT75:977 KRT6A:1248
	-0.15587105				DNMT1:74 ARID4A:86 SMCHD1:136 MAGEL2:373 PLAGL1:420 PEG10:836
MP0003121 genomic imprinting MP0002971 abnormal brown adipose	0.09686093	29 76		4.556e-02 4.556e-02	CACNA1S:152 GPR68:174 APOE:461 MOGAT2:737 RPTOR:1013 C3:1147
MP0002971 abnormal brown adipose MP0002161 abnormal fertility/fecundity		76			DLGAP5:10 CATSPER2:41 AFP:53 DNMT1:74 CLGN:84 ARID4A:86
IVICUOUZ TO L'AODOLINALIEUIIIIV/IECUDOIIV	-0.03068056	865	4.0796-03	4.854e-02	DEGARD. 10 CATORERZ.41 AFP.33 DINIVITT.74 CEGN:84 ARID4A:86

tissue_specific Top pathways by non-permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
testis	-0.13832981	1678	1.777e-68	9.598e-67	CCDC148:15 MTPAP:25 ZBBX:28 ZYG11A:34 KIF27:46 CT55:48
leg.skin	0.13887164	288	1.077e-15	2.909e-14	PKP1:8 INAVA:22 BNC1:26 DSP:30 TRIM29:35 PSAPL1:67
EBV.lymphocyte	-0.08265165	753	7.214e-14	1.298e-12	DLGAP5:10 ATAD2:27 PARPBP:29 KIF18A:30 KIF2C:108 IL23R:113
suprapubic.skin	0.12212317	259	2.103e-11	2.839e-10	PKP1:8 BNC1:26 PSAPL1:67 AADACL2:89 NKPD1:99 KRT17:105
cerebellar.hemisphere	0.07787661	542	1.300e-09	1.404e-08	SYT2:18 ADCY1:32 ARHGAP44:90 MYT1:100 PPFIA4:107 TIAM1:111
cerebral.cortex	0.14936925	110	7.057e-08	6.351e-07	ADGRB1:39 KIF1A:161 DPYSL4:262 POU6F2:425 HECW1:551 MCHR1:581
cerebellum	0.07260181	409	7.177e-07	5.234e-06	TRANK1:7 SYT2:18 GLRA4:73 MYT1:100 PPFIA4:107 TIAM1:111
skeletal.muscle	0.09025646	258	7.753e-07	5.234e-06	MYH2:11 MYBPC2:144 CASQ1:147 CACNA1S:152 MYH1:239 PYGM:292
Brodmann.area.9	0.09778598	185	5.253e-06	3.152e-05	NRCAM:96 KCNQ3:126 NECAB1:198 OLFM1:356 POU6F2:425 GPR26:479
esophagus.mucosa	0.07743980	263	1.875e-05	1.012e-04	PKP1:8 BNC1:26 ADH7:37 LAD1:42 FAM83H:124 RNF222:194
pancreas	-0.09460248	118	4.097e-04	2.011e-03	TMED6:188 CATSPERB:246 SERPINI2:302 ERP27:332 FBXW12:345 G6PC2:555
nucleus.accumbens	0.09733599	105	5.980e-04	2.484e-03	INSYN2A:113 CACNA1E:166 OTOF:272 JAKMIP1:276 SV2C:414 ARPP21:654
vagina	0.09205920	119	5.542e-04	2.484e-03	PKP1:8 BNC1:26 RNF222:194 SLURP1:253 LCE3D:304 NOD2:312
putamen	0.17511245	17	1.248e-02	4.813e-02	RGS9:52 SLC18A3:251 DIPK1C:623 OPALIN:1651 CORT:2933 LRRC10B:3207
endocervix	0.07506821	79	2.145e-02	7.723e-02	MRGPRE:1 ELFN1:41 KIF26A:66 ZNF516:93 SPON2:286 MLPH:417
greater.omentum	0.10652372	38	2.325e-02	7.848e-02	BNC1:26 AADACL4:383 ITLN1:422 THRSP:630 ADAMTS4:779 FAM89A:952
hypothalamus	0.07156423	78	2.935e-02	9.322e-02	SYT6:275 PNMA6F:477 PNMA5:688 RTL1:782 KISS1R:879 DNER:975
tibial.artery	0.05582169	113	4.124e-02	1.237e-01	SDK1:20 PPP1R12B:142 ADARB1:157 OR51E2:230 TNC:303 INHBA:349
amygdala	0.18028731	9	6.114e-02	1.572e-01	TNR:440 PTPRZ1:493 PCDH15:3256 NEUROD6:6152 SLC17A7:6846 SLC38A8:714
C1.spinal.cord	0.03971908	194	5.831e-02	1.572e-01	ACSBG1:114 CDH20:208 MAGEC3:436 CHADL:562 TTYH2:589 PLAAT3:591
sigmoid.colon	0.07755972	51	5.580e-02	1.572e-01	DES:220 HDAC4:291 TACR2:335 MRGPRD:367 NTSR1:518 GREM2:812
left.ventricle	0.06413733	68	6.810e-02	1.672e-01	MYL3:256 LMOD2:328 TNNT2:380 MYOM2:769 TENM2:1212 CKMT2:1281
peyers.patch	0.03553920	199	8.630e-02	1.970e-01	SLC17A4:9 TRPM5:13 MGAM2:118 KRT20:129 TREH:131 EPS8L3:133
transverse.colon	0.03834125	169	8.754e-02	1.970e-01	SLC17A4:9 DSG2:104 KRT20:129 EPS8L3:133 RNF186:139 MUC2:192
fallopian.tube	-0.06111042	60	1.023e-01	2.126e-01	BCHE:150 ATP8B4:268 ADAMTS3:295 CCDC17:460 TUBA4B:463 CCDC198:863
prostate	-0.06637422	52	9.841e-02	2.126e-01	CPAMD8:58 TTC6:330 SHROOM1:445 MSMB:568 TNFSF15:946 MKX:970
blood	0.02271908	347	1.515e-01	3.029e-01	HRH2:130 FFAR2:150 STX11:214 FUT7:224 TUBB1:332 PIK3R5:363
esophagogastric.junction	-0.12399366	9	1.978e-01	3.815e-01	F2RL2:1282 RTL3:2797 NKX6-1:4194 GADL1:5652 ADCY5:6799 COL4A5:7006
stomach	0.03546474	99	2.244e-01	4.040e-01	MYH13:54 PSAPL1:67 EPS8L3:133 CAPN8:171 VWA2:372 TM4SF5:379
eye.development	0.04254519	69	2.229e-01	4.040e-01	NOTCH1:258 TKT:523 BMP7:1078 TBX5:1425 CRX:1446 FGFR2:1635
hippocampus.proper	0.09538935	13	2.339e-01	4.074e-01	GRP:928 LCN1:1666 CTXND1:1693 NEUROD6:6152 OLIG2:6296 CABP7:6700
caudate.nucleus	0.06345781	28	2.456e-01	4.144e-01	GPR37L1:12 MLC1:40 TAC1:1761 PDE1B:2622 LRRC10B:3207 VAX1:3381
subcutaneous.adipose	0.03595714	82	2.618e-01	4.284e-01	GPD1:282 FASN:514 THRSP:630 F13A1:684 ACACB:896 PRG2:1036

Brodmann.area.24 0.05357897 33 2.873e-01 4.564e-01 DOK6:1564 UPP2:2270 MT3:2809 FAM189A1:2974 SHANK1:3151 GRIN3A:3627 bladder 0.05376583 30 3.086e-01 4.761e-01 UGT1A6:1295 PSCA:1571 PADI3:1835 KRT13:2204 ACSM6:2275 ABCC4:2898

3.833e-01 5.594e-01 CCDC69:740 F2RL2:1282 PRUNE2:1425 COL4A6:2457 CST5:3622 CNTNAP3:4029

NAV1:17 LGR6:51 MTUS2:243 CYP4X1:330 PDGFA:635 GABRR1:637

SLC24A4:169 ITGB4:223 ENTPD2:298 MYOC:389 INF2:410 OR6B3:501

TNN:252 THRSP:630 KRT5:1910 NPY2R:2176 PLIN1:3226 ADIPOQ:3410

transformed.skin.fibroblast 0.01656334 276 3.488e-01 5.232e-01 BNC1:26 ZNF281:81 MYOF:127 CLMP:143 MME:145 COL6A1:158

4.410e-01 6.266e-01

4.739e-01 6.561e-01

5.191e-01 6.663e-01

esophagus.muscularis.mucosa -0.04764841

aorta

tibial.nerve

breast

0.01798357

0.01462615

0.04166667

156

205

20